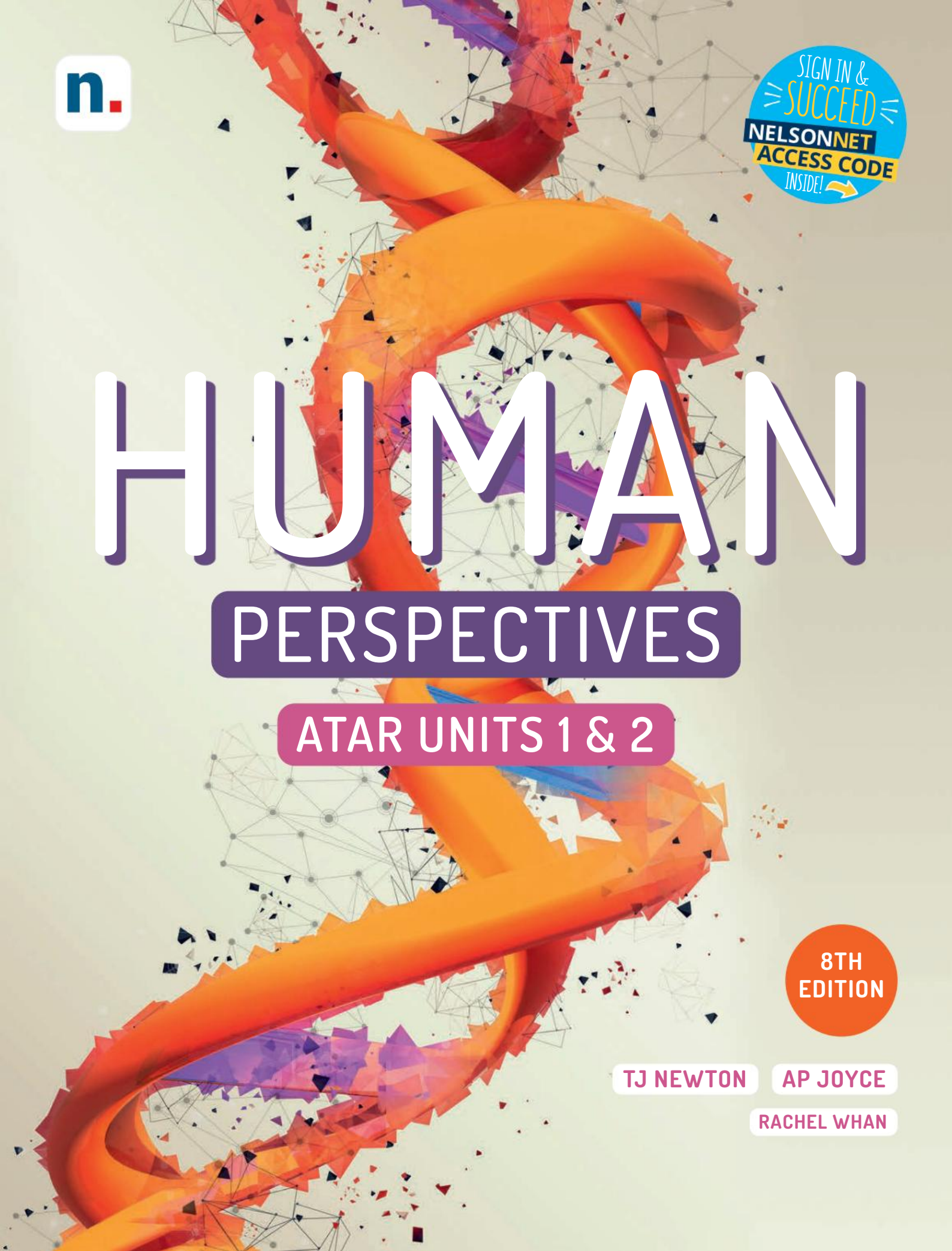
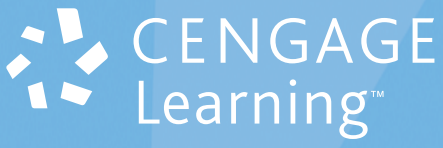

SIGN IN & SUCCEED
NELSONNET ACCESS CODE INSIDE!
HUMAN
PERSPECTIVES
ATAR UNITS 1 & 2
8TH EDITION
TJ NEWTON
AP JOYCE
RACHEL WHAN



This is a legal agreement between the you, (the "Customer") and Cengage Learning Australia Pty Limited (ABN 14 058 280 149) (the "Licensor") which provides the terms and conditions of this non-exclusive licence and the limited warranty for the Product. Use of the Product indicates an acknowledgement that the Customer has read and agreed to be bound by the terms and conditions of this Agreement. If you do not agree to these terms and conditions, return the Product to the place of purchase within 15 days of the date of purchase (with proof of purchase) for a full refund

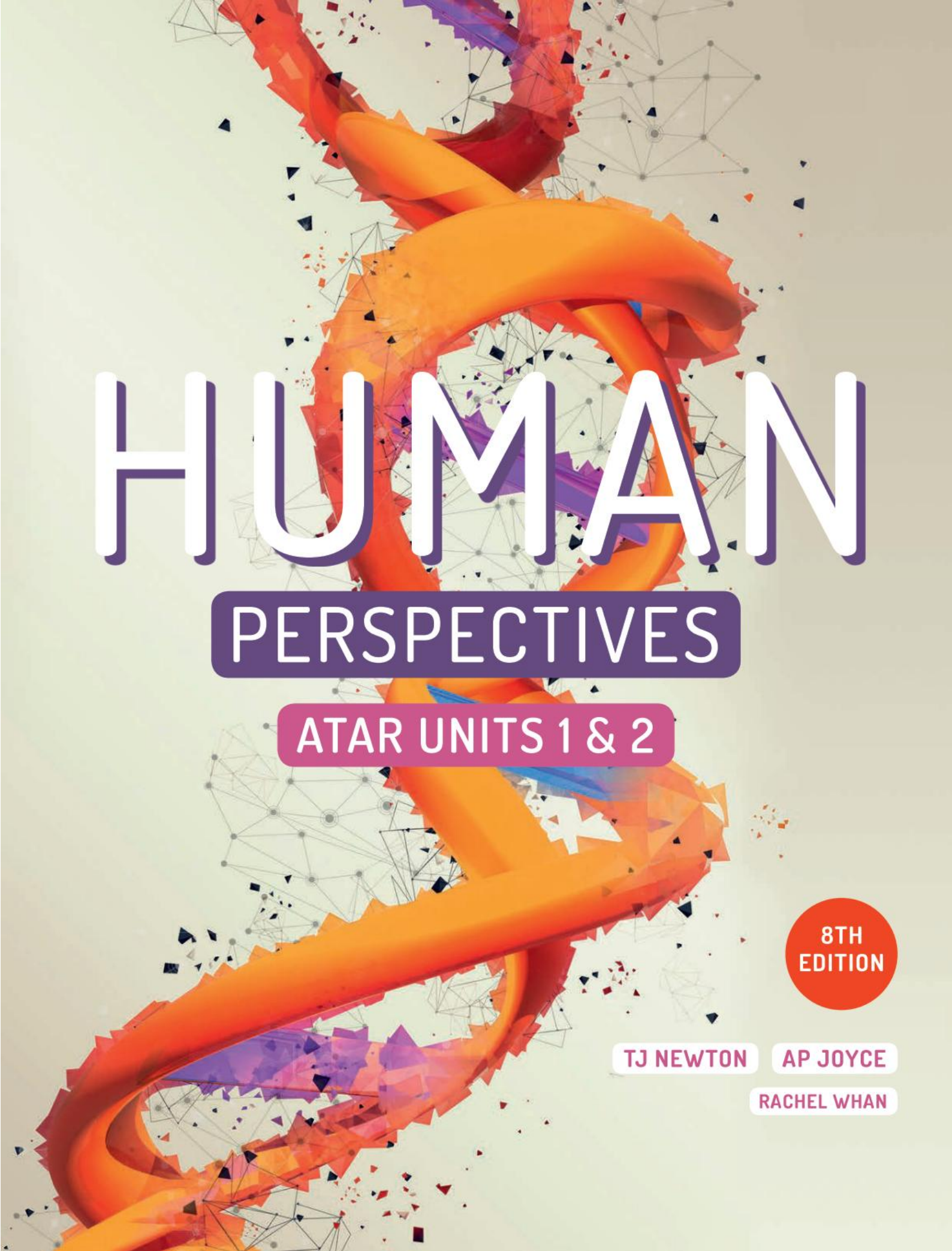
1. **Licence Grant**
   You do not receive title to the Product. Copyright in the Product (which includes all images, photographs, video, animations, audio, music and text incorporated in the Product, including all of the accompanying printed material) is owned by the Licensor and/or its suppliers and is protected by Australian copyright laws. The Licensor grants you a non-exclusive licence to use the Product subject to the restrictions and terms set out in this Agreement.

2. **A Licence allows you to:**
   Use the Product on your computer. The Customer represents that they shall in no way place the Product in the public domain or in any way compromise our copyright in the Material. You agree to take reasonable steps to protect our copyright.

3. **You may not:**
   Alter, modify, translate, reverse engineer, decompile, or adapt the software or create derivative works based on the Product. Make further copies by any means technological, electronic, digital whatsoever without the written permission of the Licensor. Rent or transfer all or any part of your rights under this Agreement. Remove or alter any copyright or other proprietary notice or label attached to the software.

4. **Termination**
   Any failure to comply with the terms and conditions of this agreement will result in the automatic termination of this licence. Upon termination of this licence for any reason, the Customer must destroy or return to the Licensor all copies of the software and accompanying documentation.

5. **Warranties**
   To the extent permitted by law, the Licensor's liability for any breach of the warranty or any term implied by law into this licence is limited to the lowest cost of replacing the goods, acquiring equivalent goods or having the goods repaired.


HUMAN
PERSPECTIVES
ATAR UNITS 1 & 2
8TH EDITION
TJ NEWTON
AP JOYCE
RACHEL WHAN

NELSON
A Cengage Company

Human Perspectives ATAR Units 1 & 2
8th Edition
TJ Newton
AP Joyce
Rachel Whan
ISBN 9780170449090

Publisher: Sarah Craig
Project editor: Robyn Beaver
Copy editor: Robyn Flemming
Text design: Rina Gargano (Alba Design)
Cover design: Jenna Lee Fai (Jenki)
Project designer: Justin Lim
Cover image: Getty Images/Victor Habbick Visions/Science Photo Library
Permissions researcher: Helen Mammides
Production controller: Alice Kane
Typeset by: SPi Global

Any URLs contained in this publication were checked for currency during the production process. Note, however, that the publisher cannot vouch for the ongoing currency of URLs.

National Library of Australia Cataloguing-in-Publication Data
A catalogue record for this work is available from the National Library of Australia.

Cengage Learning Australia
Level 7, 80 Dorcas Street
South Melbourne, Victoria Australia 3205

Cengage Learning New Zealand
Unit 4B Rosedale Office Park
331 Rosedale Road, Albany, North Shore 0632, NZ

For learning solutions, visit **cengage.com.au**

Printed in China by 1010 Printing International Limited.
1 2 3 4 5 6 7 24 23 22 21 20

CONTENTS

9780170449090

9780170449090

9780170449090

USING *HUMAN PERSPECTIVES*

Human Perspectives has been comprehensively updated to address all aspects of the School Curriculum and Standards Authority (SCSA) Human Biology ATAR course. This series will enable you, the student, to achieve maximum understanding and success in this subject. Each page has been carefully considered to provide all the information you need in a variety of different formats such as text, figures and links to online material. You will find it easy to navigate through each chapter and see connections to the practical activities and investigations through the use of icons, highlighting the importance of the interconnectedness between the conceptual and practical aspects of Human Biology.

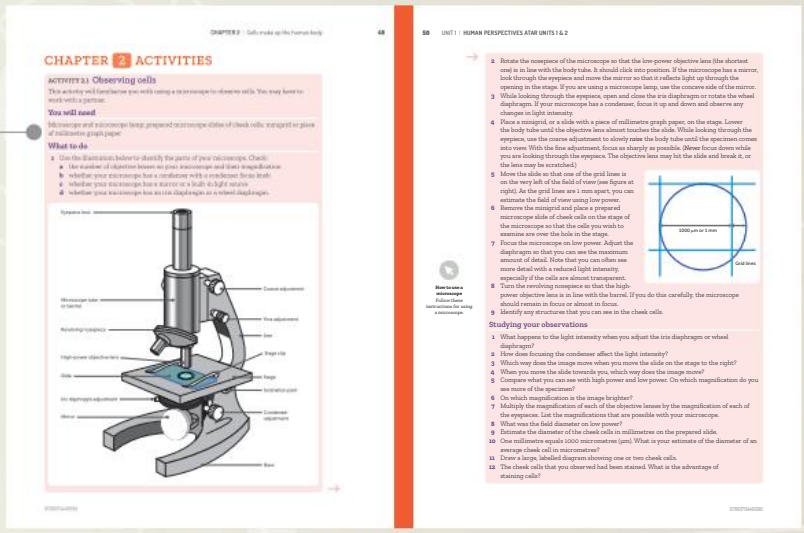
Each chapter begins with a **chapter opening page**, which presents the learning outcomes under the Science Inquiry Skills, Science as a Human Endeavour and Science Understanding strands from the SCSA Human Biology ATAR syllabus to be covered in the chapter.

Important ideas, concepts and theories are summarised in **key concept boxes** throughout chapters. These provide reinforcement and summary for improved assimilation of new ideas.

Connections to practical activities and investigations are indicated using **margin icons**. Interactive icons link to digital worksheets and websites.

Regular opportunities to recall new terms and facts, and to apply concepts, are provided in **question sets** at the end of each chapter section.

Practical activities and investigations introduce and reinforce the Science Inquiry Skills listed in the SCSA Human Biology ATAR syllabus. They allow you to explore and discover the living world.



CHAPTER 2 ACTIVITIES

ACTIVITY 2.1 Observing cells

You will need

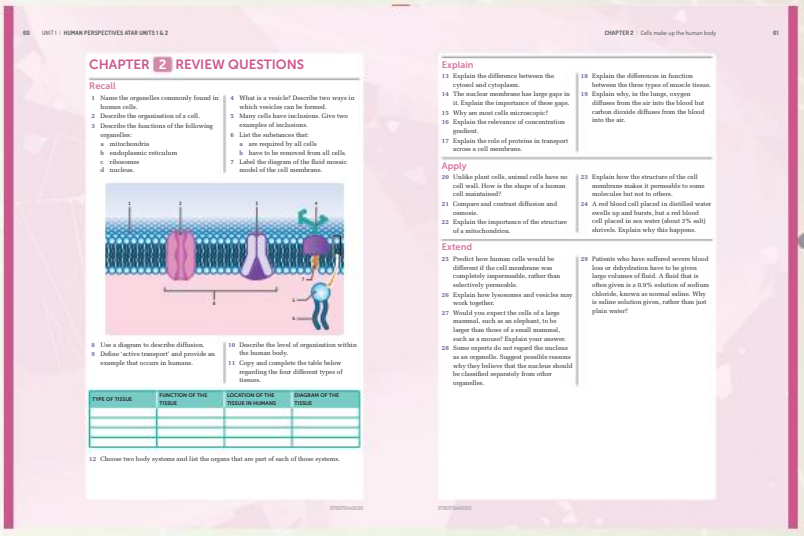
What to do

Studying your observations

Chapter review sections include a **chapter summary** for revision and reflection, a **glossary** of important terms introduced in the chapter, and **review questions** that check understanding and provide opportunities for application and analysis of concepts.

CHAPTER 2 SUMMARY

CHAPTER 2 GLOSSARY



CHAPTER 2 REVIEW QUESTIONS

Recall

Explain

Apply

Extend

PARTNERSHIP WITH SOUTHERN BIOLOGICAL

Southern Biological and Nelson Cengage have partnered to ensure that you are provided with exciting and current investigations to introduce, reinforce and practise the Science Inquiry Skills listed in the SCSA Human Biology ATAR syllabus. Some of the investigations created by Southern Biological are exclusive to Nelson Cengage, and all Southern Biological investigations have been rigorously stress-tested to ensure that they will work in your classroom.

SOUTHERN Biological Developed exclusively by Southern Biological

About Southern Biological

Southern Biological is a leading hands-on Australian science education company that has been supporting science educators for more than 40 years. With an aim to provide educators with the finest products and services to help students of all ages engage with science, Southern Biological provides an impressive range of educational resources, professional development workshops and high-quality, innovative products.

www.southernbiological.com

NELSONNET

NelsonNet is your protected portal to the premium digital resources for Nelson textbooks located at www.nelsonnet.com.au. Once your registration is complete, you will have access to comprehensive digital resources that supplement and complement each chapter, including worksheets and useful weblinks.

Icons in the NelsonNetBook link to these resources.

Teachers will have access to these resources plus answers to all student book questions and activities, practice tests, chapter PowerPoints, and syllabus mapping and teaching plan documents. Investigation support has been provided by Southern Biological and includes lab advice and safety sheets, and videos to assist both the teacher and laboratory technician in preparing and delivering the investigation to students.

PRACTICE TESTS ON COGNERO ASSESS

Practice tests for each chapter are written and presented in the style of the WACE exam (multiple-choice, short answer and extended answer questions).

They are provided on Cognero Assess, which is a powerful and flexible assessment tool for teachers. Practice tests can be modified, added to or used as is, then assigned to students for completion.

Answers are provided for all questions, with responsive marking for multiple-choice questions and marking keys for all short- and extended-answer questions. Teachers can also mark questions and review results within Cognero Assess.

NELSONNETBOOK

The NelsonNetBook is your customisable, interactive ebook that can be used online and offline. Accessible on desktops, laptops, tablets and interactive whiteboards, it reproduces the student text in digital form. Annotate your ebook using notes, highlights, weblinks and voice recordings, link to useful websites, and access resources directly from the NelsonNet student website. Teachers can use it to share their personalised version with the class or groups. You can also download an offline version of the book and iPad and Android apps.

Disclaimer

Please note that complimentary access to NelsonNet and the NelsonNetBook is only available to teachers who use the accompanying student book as a core educational resource in their classroom. Contact your Education Consultant for information about access codes and conditions.

AUTHOR ACKNOWLEDGEMENTS

A great many people have contributed to the writing and production of this book; and to all those people, the authors express their sincere thanks.

For previous editions, we are particularly indebted to June Gouldthorp and Pauline Charman. We wish to acknowledge again their vital contributions. June provided invaluable feedback on the original draft of the manuscript and helped ensure all content areas had been covered. Pauline, then Project Officer for Human Biological Sciences at the Curriculum Council of Western Australia, offered clarification of the content. We also remain grateful for the work of Audrey Sewell-Smith, Jill Lamble and Anabel Kanakis, who reviewed parts of that manuscript and provided constructive criticisms and suggestions.

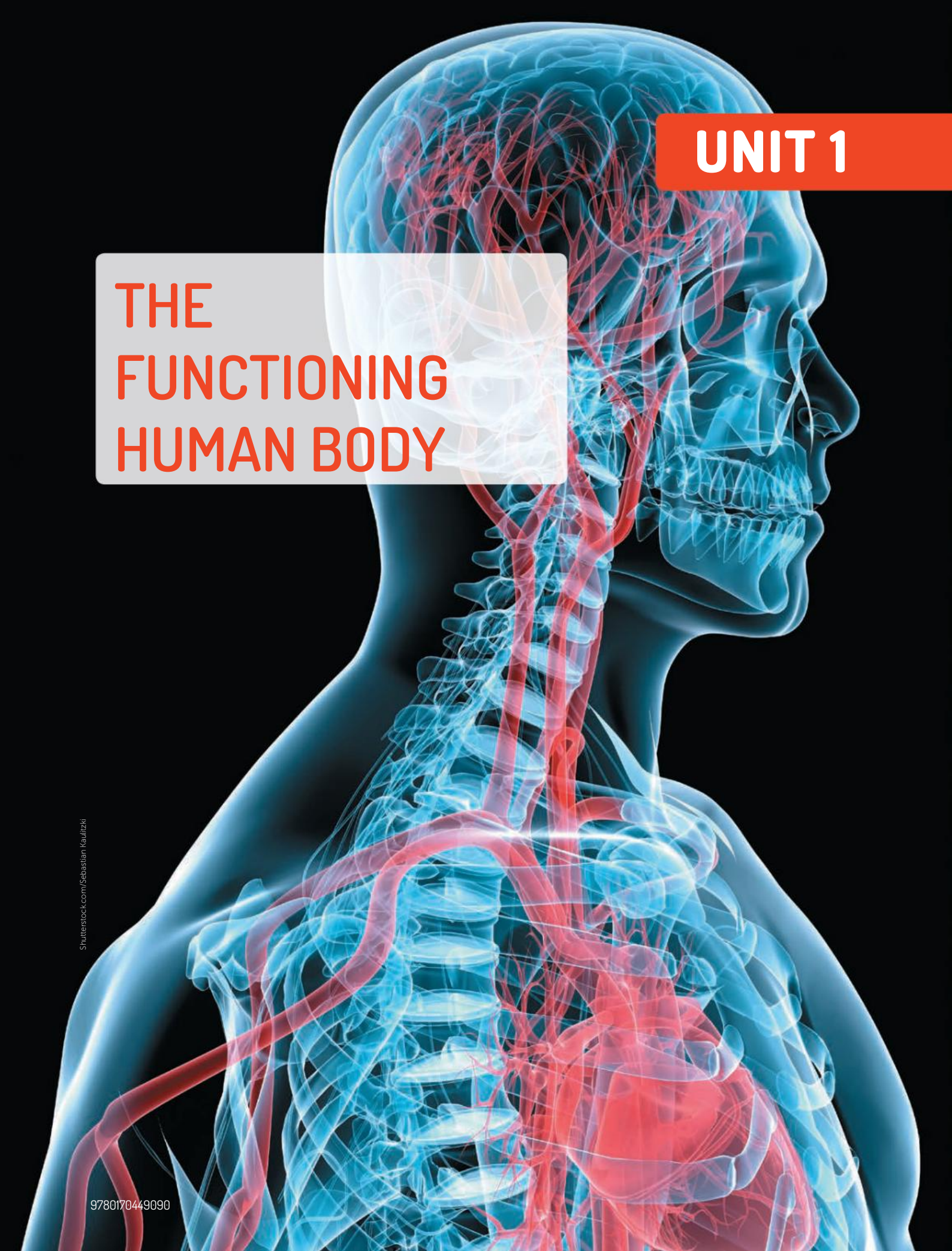
We would also like to thank those people who provided information, photographs, or both, for earlier editions. A special expression of thanks must go to Geoff Meyer of the Centre for Human Biology, University of Western Australia, for his particular contribution to past editions.

For this edition, we extend our gratitude to Derek Cumpsty, Charlotte Donovan and Michelle Moreton for their valuable feedback on the manuscript and contributions to the digital resources.

Terry Newton
Ashley Joyce
Rachel Whan

UNIT 1

THE FUNCTIONING HUMAN BODY



Shutterstock.com/Sebastian Kaulitzki

9780170449090

1 INVESTIGATING HUMAN BIOLOGY

UNIT 1 CONTENT

SCIENCE INQUIRY SKILLS

- identify, research and construct questions for investigation; propose hypotheses; and predict possible outcomes
- design investigations, including the procedure(s) to be followed, the materials required, and the type and amount of primary and/or secondary data to be collected; conduct risk assessments; and consider research ethics, including animal ethics
- conduct investigations, including monitoring body functions; use microscopy techniques; and perform real or virtual dissection, safely, competently and methodically for the collection of valid and reliable data
- represent data in meaningful and useful ways; organise and analyse data to identify trends, patterns and relationships; qualitatively describe sources of measurement error, and uncertainty and limitations in data; and select, synthesise and use evidence to make and justify conclusions
- interpret a range of scientific and media texts, and evaluate processes, claims and conclusions by considering the quality of available evidence; and use reasoning to construct scientific arguments
- communicate to specific audiences, and for specific purposes, using appropriate language, nomenclature, genres and modes, including scientific reports

Source: School Curriculum and Standards Authority, Government of Western Australia

9780170449090

1.1 STUDYING HUMAN BIOLOGICAL SCIENCE

Human biological science is the scientific study of humans, both as individuals and as populations, and the study of the interaction between humans and their environment. Although a lot is now known and understood about humans, much still remains to be discovered. A glance through almost any newspaper will show that discoveries are continually being made about how our bodies work, about human behaviour and about where we came from. Despite the progress in knowledge of our species, new problems constantly arise.

What is science?

There are two aspects to science.

- Science is a process of inquiry – a way of finding out about human beings, and their living and non-living surroundings.
- Science is a body of knowledge – knowledge gained by systematic observation and testing of ideas.

Because of the nature of science as a process of investigation, scientific knowledge is not fixed and unchanging. Knowledge is continually increasing as new discoveries are made, and existing knowledge often has to be modified, or even discarded, as new evidence accumulates.

What is human biological science?

Human biology is a body of knowledge relating to humans and is concerned with finding out more about the human species. This book presents some of the knowledge that scientists have gained about humans, sometimes with a description of how that knowledge was gained. It is important to remember when reading this book, or any science textbook, that the information was originally gained by scientific investigation, and that the ideas presented are subject to change as research reveals more about the topics covered.

Because human biology is a science, our knowledge of humans builds on the discoveries made by each generation of scientists. Discoveries made today become the building blocks for the knowledge of tomorrow. Collective knowledge of the human organism is now so great that human biologists must specialise in a particular field of study relating to humans, such as anatomy, physiology, nutrition or biochemistry. Some of the important fields of study that contribute to an understanding of our species are shown in Figure 1.1 and Table 1.1. Aspects of many of these fields will be discussed in this book.

Key concept

Human biology is an area of science that studies humans and their interactions. It utilises a wide range of fields of study to develop an in-depth knowledge of humans.

How do scientists investigate?

How are discoveries made? What procedures does a scientist use to gather new information? There are no hard and fast rules, and no particular sequence of steps that must be followed. Different scientists investigate in different ways; however, in every case, scientific investigation begins with a problem – a question that needs to be answered. Curiosity is a human characteristic. We like to know how and why things happen. The scientist defines a problem and then tries to find a solution to that problem. Some of the ways that scientists investigate are described in the following pages.

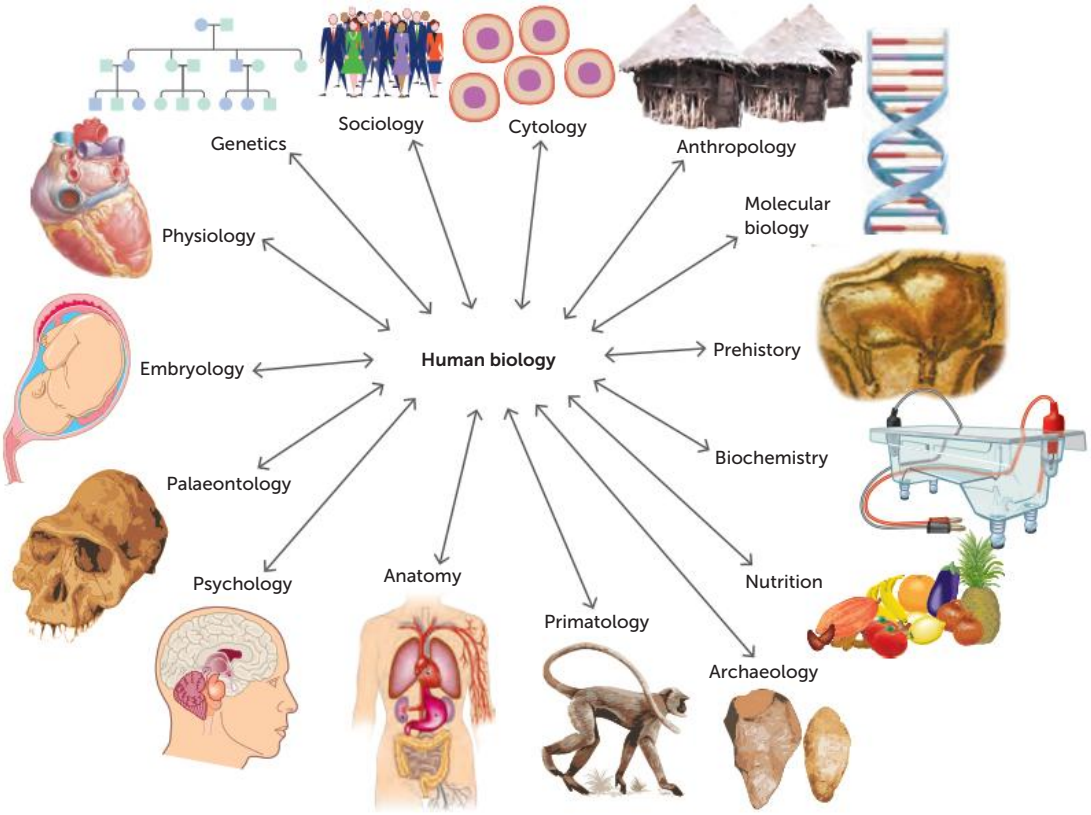


FIGURE 1.1 Some of the fields of study that contribute to our understanding of the biology of humans

TABLE 1.1 Some important fields of study that contribute to our understanding of the biology of humans

FIELD	AREA OF STUDY
Anatomy	Structure of the body
Anthropology	Relationships between the biological, cultural, geographical and historical aspects of humans (some people use the word 'anthropology' to mean the same as human biology, although human biology covers a wider field)
Archaeology	Material evidence of the past such as tools, weapons and art, rather than written records
Biochemistry	Chemistry of living things
Cytology	Cells
Demography	Statistical study of populations
Embryology	Development from fertilisation to birth
Genetics	How characteristics are passed from generation to generation
Molecular biology	Macromolecules of the cell
Nutrition	Food requirements of humans
Palaeontology	Fossils
Physiology	Functioning of living things
Prehistory	The past, before the time of written records
Primatology	Non-human members of the order Primates – apes, monkeys, lemurs, lorises and tarsiers
Psychology	Human behaviour
Sociology	Nature of human society

9780170449090

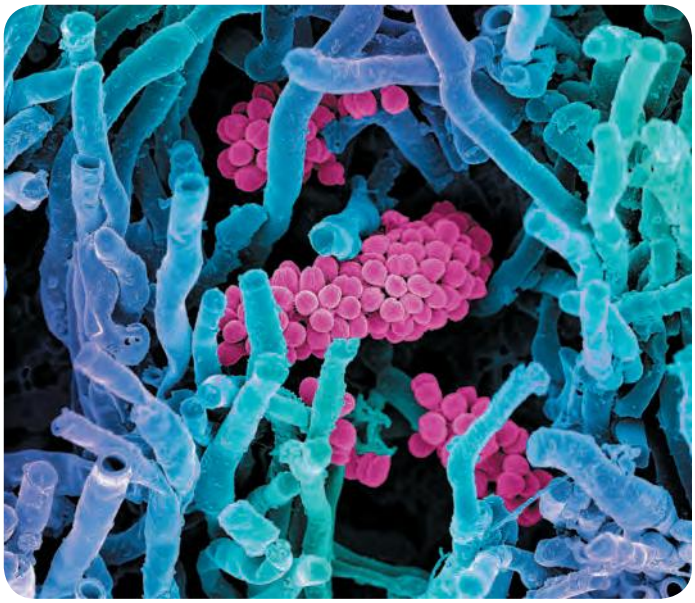
Literature review

The search for a solution to a problem will almost certainly involve a review of books, scientific journals and the Internet to see what information has already been collected by others. In this way, science builds on past discoveries and the scientist does not duplicate work already done by other scientists.

Observation

Some problems can be solved by **observation**. Information is gathered using the senses or instruments that enhance the senses, such as a microscope or stethoscope. For example, scientists who study monkeys and apes, close relatives of humans, will spend much of their time observing and noting what the animals do and how they behave towards one another. Microbiologists observe the structure of bacteria and viruses using electron microscopes. Archaeologists excavate ancient human living-sites and observe the type and distribution of shelters, tools, weapons, animal bones and charcoal from fires. Observation in the field is then followed by detailed laboratory examination of the material collected.

Patient observation produces data that must then be carefully analysed. Scientists look for patterns or trends in the data and try to draw meaningful conclusions. Often the observations will raise many new questions to be answered and problems to be solved.



Shutterstock.com/Kateryna Kon

FIGURE 1.2 Bacteria are observed, using a scanning electron microscope

Classification

Classification, placing things in groups based on the similarity of their characteristics, is a basic part of science. For example, astronomers classify galaxies by their shape and planets by their composition; geologists classify rocks according to the way they were formed and the minerals they contain; chemists classify substances by their properties; and biologists use structural features as a basis for classifying living things.

Some biologists specialise in the classification and naming of organisms, but all use classification from time to time. The classification of animals is based mainly on structural characteristics. It highlights the similarities and differences between groups and makes communication easier. For example, if we say that a particular animal is classified as a mammal, we do not have to be given a description of its characteristics. We immediately know that it has hair, is warm-blooded and suckles its young on milk. We can then think of many other animals with those same characteristics to which it is related. Classification, therefore, serves to facilitate scientific study.

9780170449090

Human biologists are concerned with the classification of the Primates, the order of mammals to which humans, apes, monkeys and some other animals belong. Although based on structural features, there is sometimes disagreement about the relative importance of certain structures. Even today, debate is continuing about how groups within the order Primates should be classified. Now that it is possible to compare the DNA of different species, biologists can be more certain about the relationships between groups. However, extinct animals that are known only through fossils may be very difficult to classify and may provoke much controversy among experts.

Experimentation

Many scientists conduct experiments. They will propose a *hypothesis*, a possible explanation or solution to a problem, and then design experiments to test it. An experiment must be designed so that the results clearly support or disprove the hypothesis being tested. To achieve this, only one factor, or **variable**, is tested at a time. A **control** test is conducted using standard conditions and an experimental test is conducted with one variable changed. If a scientist were testing the hypothesis that bacteria are killed by household bleach, two groups of bacterial cultures would be needed: experimental cultures and control cultures. The experimental cultures would be exposed to household bleach, whereas the control cultures would be exposed to distilled water. All other variables, such as the nature of the culture medium, the size of the culture containers, the type of bacteria, the temperature at which the cultures were kept, the length of time for which the cultures were observed and the volume of liquid added to each culture (bleach or distilled water), would have to be the same for both groups. These are known as *controlled variables*. If, at the end of the experiment, the bacteria in the experimental cultures were dead, whereas those in the control cultures were living, one could confidently say that the difference between the two sets of bacteria was due to the presence or absence of household bleach. This interpretation could only be made if just *one* variable was allowed to differ between the two groups of bacterial cultures.

Shutterstock.com/grebcha

FIGURE 1.3 Bacterial cultures can be compared to determine the effect of household bleach on bacteria

Key concept

Scientists use literature reviews, observation, classification and experimentation to learn more about humans and to extend our knowledge.

Questions 1.1

RECALL KNOWLEDGE

1. List the two aspects of science.
2. Define 'human biological science'.
3. Define 'psychology', 'biochemistry' and 'cytology'.
4. List the methods of investigating.

APPLY KNOWLEDGE

5. Explain how physiology relies on a knowledge of anatomy.
6. Give an example of where observations are used during scientific investigations.
7. Explain why a control experiment is an important part of a scientific investigation.

1.2 SCIENTIFIC METHOD

How are discoveries made? What procedures does a scientist use to gather new information? There are no hard and fast rules, and no particular sequence of steps that must be followed, but scientific investigation usually follows a pattern that is known as the **scientific method**. The precise method will be unique to the particular circumstances, but the underlying pattern of logical thought is similar in all cases.

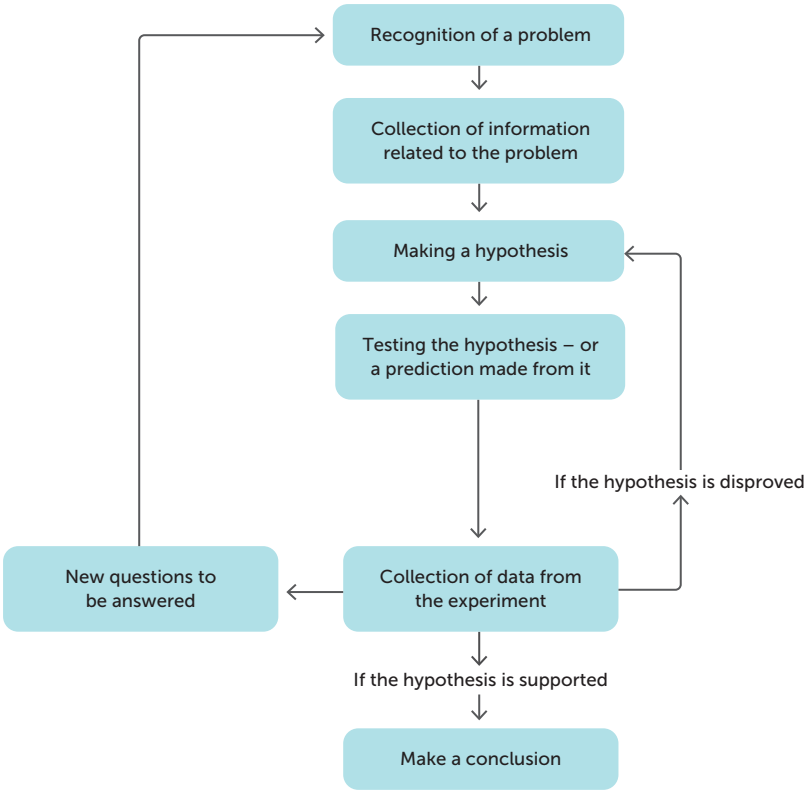


FIGURE 1.4 The scientific method

Identifying a problem

Scientific investigation begins with a problem. Curiosity about why and how things happen is a characteristic of human nature. It raises all sorts of questions to be answered and problems to be solved. Sometimes it is difficult to know the right question to ask, but until someone asks the question, research into the answer cannot begin. For example, Louis Pasteur (1822–95) was curious about the cause of infectious disease and of fermentation. In investigating these problems, he found that micro-organisms are responsible for many diseases and for fermentation. Knowing what questions to ask is one of the characteristics of a good scientist.

Collecting information

Having defined the problem, the scientist begins to collect information about the problem. This may involve direct observation. For Pasteur, this was looking at samples of fermenting liquid to see if they contained micro-organisms. It will almost certainly involve a review of books, scientific journals and the Internet to see what information relating to the problem other researchers have collected. In this way, science builds on past discoveries and the scientist does not duplicate work already done by other scientists.

Identifying variables

A variable is any factor that may change during an experiment. In an investigation, variables are classified as either independent, dependent or controlled.

- The **independent variable** is the factor that is being investigated. This means that it is the factor that is deliberately changed to determine how it affects the results. The independent variable may also be called the experimental variable or the manipulated variable.
- The **dependent variable** is the factor that changes due to the changes made to the independent variable. It is sometimes called the responding variable.
- **Controlled variables** are the factors that are kept the same for both the control and experimental groups in an experiment.

Developing a hypothesis

After collecting information related to the problem, the scientist can then suggest an explanation for the observations. This is called a **hypothesis**. For example, Louis Pasteur observed that certain diseases seemed to be passed on from one person to another. This gave rise to the question of what caused these diseases. One of Pasteur's hypotheses was that micro-organisms in the air were the cause.

A hypothesis requires investigation to collect evidence that will support it, and therefore must be testable. A good hypothesis:

- is a definite statement, not a question
- is short (it is much easier to test a simple hypothesis than a complex one)
- has a single idea that can be tested
- usually links the independent and dependent variables – for example, Pasteur's hypothesis that micro-organisms (one variable) cause disease (second variable). In most cases, the link will be in the form of a trend.

Note that a hypothesis must be able to be tested. The belief that individual species were created by God, or that hip hop is better than classical music, are not hypotheses because they cannot be tested. Science cannot test matters of religious faith or personal taste in music.

9780170449090

A hypothesis should state what you think the relationship is between the variables. For example, a student might be interested in a possible link between sweating and urine production. Three hypotheses are possible:

1 'Sweating causes a decrease in urine production.'
2 'Sweating increases urine production.'
3 'Sweating has no effect on urine production.'

These are all valid hypotheses because they state the relationship between the two variables: sweating and urine production. 'Sweating affects urine production' would not be a good hypothesis, because it does not specify the relationship between the two variables.

Having made a hypothesis, the scientist often makes a **prediction** of the results based on the hypothesis. For example, if the hypothesis was 'sweating causes a decrease in urine production', then a prediction may be that urine production will be least in people who sweat the most.

Key concept

A hypothesis is a statement of the expected relationship between the independent and dependent variable that can be tested.

Testing the hypothesis

The next step is to **test** the hypothesis, or a prediction made from the hypothesis, by using a suitable experiment.

To ensure that the results of an experiment will either support or disprove the hypothesis, only one factor, or variable, is tested at a time. A control, or comparison, experiment is also done in which nothing has been changed. If a scientist were testing the hypothesis that vitamin A is essential for the normal development of young rats, two groups of young rats would be needed: an experimental group and a control group. The experimental group would be given a diet deficient in vitamin A, while the control group's diet would contain normal amounts of vitamin A. All other variables, the controlled variables, are the same for both groups – for example, the age and sex of the rats, the type and quantity of the food, the length of time for which the rats were observed, and the temperature under which the rats were kept. If, at the end of the experiment, the experimental rats were small and underweight for their age, whereas the control rats were of normal size and weight, one could confidently say that the difference between the two sets of rats was due to the presence or absence of vitamin A in their diet. This interpretation could only be made if just *one* variable were allowed to differ between the two groups of rats. Such an experiment, involving an experimental group and an appropriate control group, is known as a **fair test**.

During the experiment, the scientist observes and records all the information from the experiment. This information is called the **data**. Whenever possible, the results of an experiment should involve measurement, or **quantitative data**, and direct observation, or **qualitative data**. A scientist would record the appearance of the rats in this imaginary experiment, but more importantly would measure things such as the mass of the rats and the length of the rats' bodies or body parts. Measurement is precise and is easier to compare than descriptions of observations.

Scientific experiments always involve **repetition**. This may mean doing the same experiment many times, or it may mean performing the experiment on a large number of subjects at the same time. In the rat experiment, one experimental rat and one control rat would not be sufficient as there is natural variation within any species. One of the rats may be slightly unusual or abnormal in some way. (This would be an uncontrolled variable.) If this were so, the result could lead the scientist to the wrong interpretation. If 10 experimental and 10 control rats were used, the average change in weight over the period of the experiment could be calculated. Any chance differences between individual rats would then be unlikely to affect the result.

Activity 1.1
Hypothesising

Activity 1.2
Investigating how pollen causes hay fever

Activity 1.3
Designing controlled experiments

Activity 1.4
Testing a hypothesis

Presentation of data

The aim of the investigation is to identify the relationship between the independent and dependent variable. In order to see any trend, the data needs to be presented in an organised manner. Tables and graphs are effective tools to organise data.

Tables

Results of investigations are often presented in the form of a table. A table is an organised and concise way of presenting data. Observations may be presented in this form; however, tables are particularly useful for presenting numerical data.

When you draw up a table to present the results of an experiment, there are certain rules you should follow:

- The table must have an informative title. The title usually states the variables investigated in the experiment.
- Data is presented in columns. Usually, the data for the independent variable is in the left column, and data for the dependent variable is in the right column or columns. However, this is not a definite rule. The most important consideration is that the table is easy to understand.
- Each column has a heading that names the variable and the units in which it is measured.

For example, the results of the experiment on the effect of vitamin A on the growth of young rats could have been presented in the way shown in Table 1.2. The independent variable, type of diet, is shown in the left-hand column, and the dependent variable, body weight, is shown in the right-hand columns.

TABLE 1.2 The effect of vitamin A deficiency on the body weight of rats

TYPE OF DIET	BODY WEIGHT OF EACH RAT AT THE END OF THE EXPERIMENT (g)										
	1	2	3	4	5	6	7	8	9	10	AVERAGE WEIGHT
Normal diet											
Diet deficient in vitamin A											

Graphs

A useful way to present data so that it can be understood easily is to draw a graph. A graph shows how changes in one variable affect a second variable. For example, if the weight of a baby is measured every month for two years, the data can be plotted on a graph. Time (in months) is one variable; it affects the other variable, weight. In this case, time is called the independent variable. Weight is the dependent variable because the weight of the baby depends on the month in which it was measured (the month does not depend on the weight of the baby). The independent variable is normally plotted on the horizontal axis of a graph and the dependent variable on the vertical axis. Such a graph would look like Figure 1.5.

When drawing a graph:

- Include a title that summarises the relationship between the variables as illustrated by the graph. This is usually written below the graph, as graphs are classified as figures.
- Label the axes with the names of the variables.
- Indicate the units in which each variable is measured.
- Use a scale with equal intervals of units on each axis.

Activity 1.5
Testing the product claims for Hairnu

Activity 1.6
Tabulating data

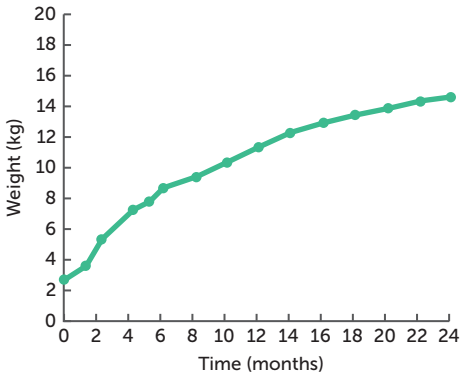
Line of best fit
This website has more information about how to draw a line of best fit.

9780170449090

Line graphs

Figure 1.5 is a **line graph**, the most commonly used type of graph in science. Line graphs are used when **continuous data** is being represented. Measurements such as length, mass, time, pH and volume are all examples of continuous data.

When there is a trend between the variables, a line of best fit is drawn, rather than joining the individual points. This may be a straight line or a smooth curve.

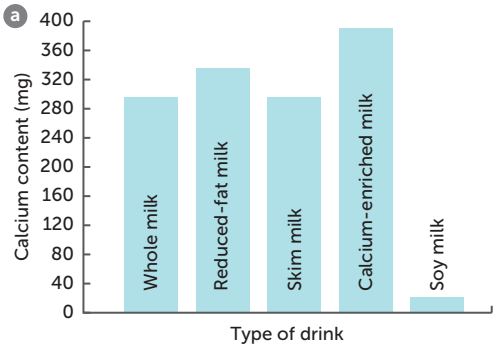


Graph 1 The weight of a baby over two years

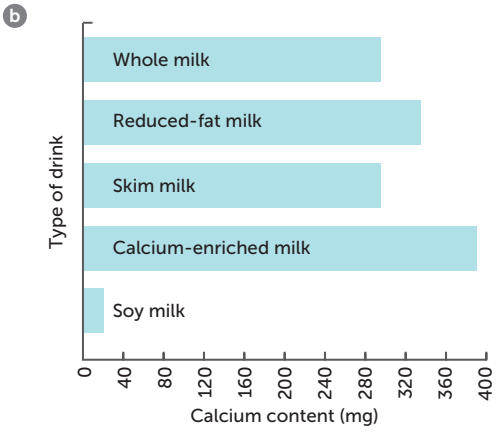
FIGURE 1.5 A line graph

Bar and column graphs

If discrete data is being graphed, a **bar** or **column graph** is used. In these graphs, the data is represented by rectangles of equal width, with spaces between them. The length of each rectangle indicates the quantity, and so the various quantities can be compared easily. Rectangles are drawn horizontally for a bar graph and vertically for a column graph. Figure 1.6 shows a column graph and a bar graph of the amount of calcium in soy milk and different types of milk.



Graph 2 Milligrams of calcium in a 250 mL glass of milk or soy milk



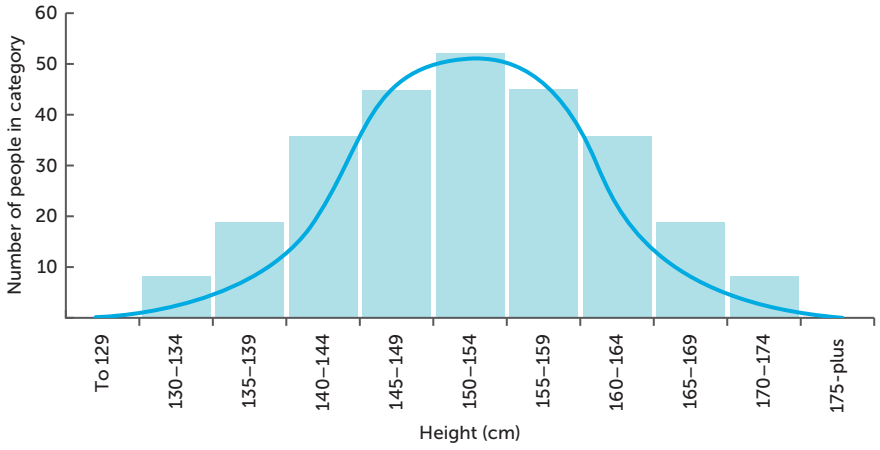
Graph 3 Milligrams of calcium in a 250 mL glass of milk or soy milk

FIGURE 1.6
a Column graph
b Bar graph

Activity 1.7
Graphing

Histograms

Sometimes data is better represented by a **histogram**. Histograms are often used to show frequencies, which is how often a particular value or characteristic occurs. They have columns to represent the frequency. The columns are of equal width, but there is no space between them. Histograms are used particularly where the data has been grouped into categories to make it more manageable. For example, a histogram would be used to graph the number of Year 12 students with heights in the categories of 130–134 cm, 135–139 cm, 140–144 cm, and so on.



Graph 4 The height of students in Year 12

FIGURE 1.7
Histogram

9780170449090

Key concept

Tables and graphs are used to organise and display data in order to identify trends.

Interpreting the data

After all the data is collected, the scientist can make an **interpretation** of the results. Data from a well-designed experiment will either support or disprove the hypothesis. If the hypothesis is disproved, the scientist must make a new one or modify the original hypothesis. If the scientist concludes that the hypothesis is supported, they communicate the **conclusion** to others in a report with details of the experiments and the data collected. It is important to note that the results of an experiment cannot prove a hypothesis: a hypothesis can only be supported or disproved.

Eventually, if enough supporting evidence is collected and there is no evidence against the hypothesis, then the hypothesis will become a **theory**. Examples of scientific theories that have been widely accepted because they have a huge amount of evidence to support them are the atomic theory, Einstein's theory of relativity, and Darwin's theory of evolution through natural selection. The use of the word 'theory' in science is different from the everyday use of the word. A scientific theory has been established and verified through investigation. It has been accepted as valid because it has been repeatedly tested.

Evaluating the experiment

As scientists, it is important that we reflect on both the results and the method. In particular, we should consider the following:

- The **validity** of the method, and therefore of the conclusion: a valid method will fairly test the hypothesis so that the only factor that affects the results is the independent variable. This means that we can trust the conclusion from the data.
- The **accuracy** of the results: this refers to how correct or true the measurements are. Accurate data is possible if the correct measuring equipment is used.
- The **reliability** of the results: this refers to how similar the results for trials of the same test are. We are able to identify reliable results by conducting multiple trials and comparing the measurements. If the method is followed correctly each time, the variables are controlled, and the equipment used is appropriate, then the reliability of the results is increased.

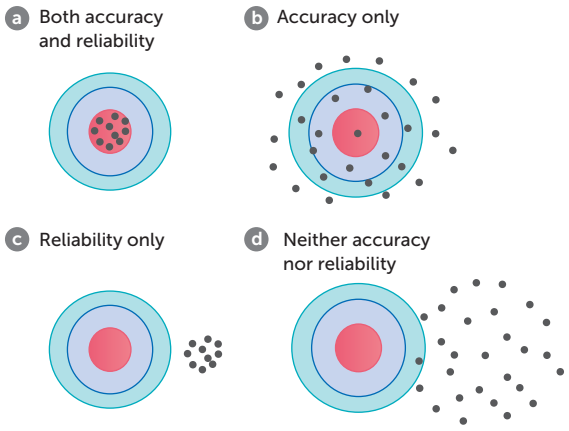


FIGURE 1.8 The difference between 'accuracy' and 'reliability'

Key concept

Investigations can be evaluated in terms of their accuracy, reliability and validity.

Experimental error

Results of experiments always contain errors. This is one of the reasons why scientists rarely make definite statements about their results. Rather than make an exact statement, they are likely to say: 'It is probable that ...' or 'It is likely that ...'. Experimental error is also one of the reasons why favourable experimental results cannot prove a hypothesis. They can only provide support for it.

In designing experiments, it is important to be aware of possible sources of error and to minimise them as far as possible.

There are three possible types of error that may occur in an experiment.

- A **human error** is simply a mistake – for example, incorrectly reading the scale on an instrument, spilling some liquid before measuring the volume, or making a mistake in a calculation. Human errors are not part of experimental error. They should be avoidable with sufficient care and checking.
- **Random errors** are unpredictable errors that can occur in all experiments. They occur because no measurement can be made with absolute precision. For example, if you are using a stopwatch to time how long it takes a person to carry out a particular task, sometimes you will stop the watch a little early, sometimes a little late. This is not human error; it is a limitation of the timing procedure. Because such an error is random, the impact of the error can be reduced by taking several measurements and averaging them.
- **Systematic errors** occur because of the way in which an experiment is designed or due to problems with equipment. In this case, a measurement will always be too high or too low. Systematic errors cannot be reduced by averaging; the only solution is to change the experimental procedure or equipment.

Scientific method

Key concept

Errors in experiments may be classified as human error, systematic error or random error.

1.1 Investigating

Questions 1.2

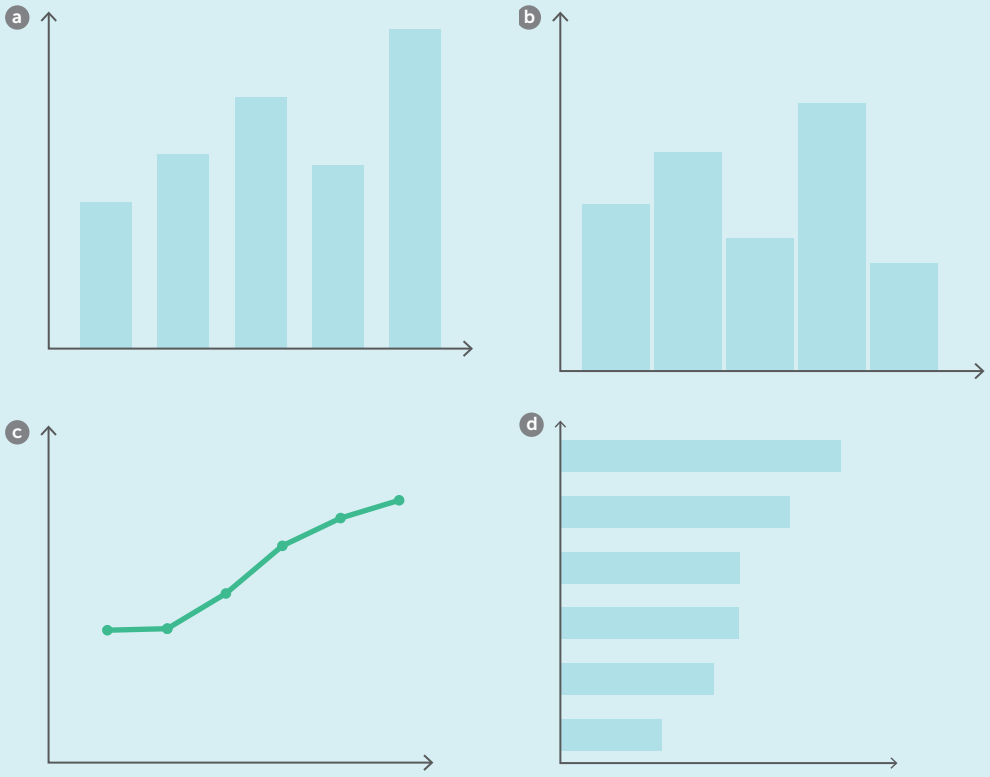
RECALL KNOWLEDGE

1 List the steps in the scientific method.
2 Define 'independent variable', 'dependent variable' and 'controlled variable'.
3 Is a baby's birth weight quantitative or qualitative? Justify your answer.
4 Which variable should be on the horizontal axis of a graph?
5 Define 'validity', 'random error', 'conclusion', 'reliability', 'systematic error', 'accuracy' and 'theory'.

APPLY KNOWLEDGE

6 A scientist was testing the effect of caffeine on memory.
  a What is the independent variable?
  b What is the dependent variable?
  c Write a hypothesis for the test.
7 Explain why it is important to collect information in the early stages of a scientific investigation.
8 Suggest why a hypothesis should be about a single idea.
9 The hypothesis for an investigation is: 'The darker a person's skin colour, the less likely they are to develop skin cancer.'
  a State the independent variable.
  b State the dependent variable.
  c List five factors that need to be controlled for the investigation to be valid.
  d Explain why it is important that a large number of people are included in the investigation.
10 What type of graph should be used to represent data about the number of people with different eye colours?

11 Classify each of the following graphs as either a column graph, bar graph, line graph or histogram.



12 'Systematic errors may produce reliable results.' Discuss whether this statement is true or not.

1.3 INVESTIGATING HUMANS

There are some additional considerations when conducting investigations on animals, including humans.

Ethical problems

Ethics are a set of moral principles or values; **ethical behaviour** is behaviour that conforms to those principles or values. In scientific research, particularly research with human participants, many ethical issues arise.

Every Australian university or research institute is required to have an ethics committee. The committee members represent a wide variety of interests and must include some members who have no links to the university or institution. Ethics committees examine proposals for research involving humans and, if the proposed investigation satisfies ethical standards, give approval to go ahead.

The following are some of the principles that an investigation must satisfy if it is to be ethically sound:

- *Voluntary participation:* people should not be pressured into taking part in the research.
- *Informed consent:* the participants should be fully informed about the objectives of the research, the procedures to be followed, any possible risks and the potential benefits; consent (in writing) should only be sought after all information has been given.
- *Risk of harm:* for some research, such as testing new drugs, it is difficult to ensure that there is no risk that participants will be harmed, but the possibility of harm should be minimised and the relationship between the risk and the benefit should be carefully assessed.

9780170449090

- *Confidentiality:* procedures need to be adopted to ensure that the identities of participants will not be revealed except to people directly involved in the study.
- *Anonymity:* this is a stronger guarantee of privacy than confidentiality; the participants in the study remain anonymous, even to the researchers. Because of the nature of some research, anonymity may not be possible – for example, where measurements must be made on participants over an extended period of time.

An ethical dilemma may arise when the effects of a trial on the experimental group of subjects are so advantageous that it seems unfair to withhold them from the control group. For example, if a new medical treatment for a disease was tested on an experimental group, which showed remarkable improvement compared to a control group, why should the control group be denied access to the treatment? Situations have actually occurred where the testing of a procedure has been so successful that the trial has been abandoned and the procedure has been made available to the control group and the experimental subjects. An example of this was a 2005 study of 3000 men living in a township in South Africa (*New Scientist*, 25 November 2006, p. 8). Volunteers who were circumcised at the beginning of the study were found to be 60% less likely to become infected with human immunodeficiency virus (HIV) than the control group of men who remained uncircumcised. The results were so dramatic that the trial was halted early so that the uncircumcised men could be circumcised.

Ethical problems may also arise when subjects are being adversely affected by the research. At what point should the trial be abandoned, even if continued testing is desirable?

Investigations that you will carry out in your human biological science course are unlikely to raise any serious ethical problems. However, it is a good idea to keep the five principles described above in mind when designing an investigation.

Key concept

Investigations involving humans must be assessed with regard to ethical considerations prior to commencing.

Placebos

Placebos are used in research into the effectiveness of medical treatments, such as a new medicinal drug. In the case of a drug trial, a **placebo** is an inactive substance that looks like the real medication. One group of subjects, the experimental group, takes the drug that is being tested and the other group, the control, takes the placebo. The placebo should look exactly the same, and be given in the same way, as the drug being tested. Subjects do not know whether they are receiving the drug or the placebo. If there is a clear difference in results between the new drug and the placebo, then the researcher can say with confidence that the difference was due to the effectiveness of the drug.

A placebo does not have to be a tablet. It could be any 'dummy' treatment, such as an injection, a skin patch, a nasal spray, a special diet, a physical therapy or even mock surgery. The important thing is that the subject believes that they are receiving exactly the same treatment as everyone else in the trial.

Patients who are given a placebo often show an improvement in their condition even though the placebo is inactive. This is called the **placebo effect**. It is thought to occur because of the patient's belief that the placebo is a real therapy that will bring about improvement. Experiments involving placebos are usually **blind experiments** where the subjects do not know whether they are receiving the placebo or the treatment. In a **double-blind experiment**, neither the researcher nor the subjects know who is receiving the treatment or the placebo. These precautions reduce the risk of any bias due to the placebo effect. If, in a trial of a therapy, the test group shows a better response than the control group, despite the placebo effect, the therapy can be assumed to have been effective.

Questions 1.3

RECALL KNOWLEDGE

1. Define 'ethics'.
2. List the principles that must be satisfied for an ethically sound investigation.
3. Describe a double-blind experiment.

APPLY KNOWLEDGE

4. Explain the difference between confidentiality and anonymity.
5. Explain why placebos are an important part of medical research.
6. During research into a new treatment for breast cancer, one group of patients took a new drug while another group took a placebo under the same regime. The patients in the placebo group showed a slight improvement, while the patients taking the drugs showed signs of kidney failure. Discuss whether or not the research should be continued.

9780170449090

CHAPTER 1 ACTIVITIES

ACTIVITY 1.1 Hypothesising

In 1876, Robert Koch, a German doctor, was the first person to demonstrate that a particular type of bacterium caused a specific disease. He showed that a rod-shaped bacterium, later called *Bacillus anthracis*, caused anthrax, a disease that occurs in sheep, cattle, horses and sometimes humans.

Which of the following could have been suitable hypotheses for Koch to investigate in his search for the cause of anthrax? For each of the items below, give reasons why it would or would not be a suitable hypothesis.

1. *Bacillus anthracis* causes anthrax.
2. Can anthrax be passed from sheep to cattle?
3. If a sheep is injected with *Bacillus anthracis*, it will get anthrax.
4. To look for *Bacillus anthracis* in the blood of animals with anthrax.
5. Why does *Bacillus anthracis* cause anthrax?
6. If a cow is injected with *Bacillus anthracis* and is then kept out of the weather, it will not get anthrax.
7. Injecting blood from a sheep suffering from anthrax into a healthy sheep will transmit the disease.
8. Any animal suffering from anthrax will have *Bacillus anthracis* in its blood and will pass the infection on to other animals.

ACTIVITY 1.2 Investigating how pollen causes hay fever

Read the following newspaper article extract carefully and then answer the questions below.

> For years scientists were puzzled at how a grain of pollen that was too big to get past the defences of the nose and throat and into the lungs could trigger breathing problems from allergy and asthma.
>
> Murdoch University environmental scientist, Frank Murray, said it was known that only airborne objects with a diameter of 10 microns or smaller (one micron is one-thousandth of a millimetre) could pass through into these vital organs, yet grass pollens measured up at 20 to 30 microns.
>
> But with the aid of the magnification of a powerful microscope, they have recently discovered that grass pollen swells and bursts when in contact with water, splitting like a damaged can of baked beans.
>
> 'We saw a big particle but with smaller, little granules running out of it,' Professor Murray said. He now believes the same reaction occurs when it rains on grass pollen or if the pollen hits a moist membrane in the body such as the nose, eyes or throat.
>
> 'This is only one possible mechanism. But if they break up to particles only one micron in diameter, then it is easy to see how they get into the lungs.'

9780170449090

Professor Murray said this could partly explain why some people complained of hay fever following rainfall, though rain was also known to trigger the mass release of fungal spores to which some people were highly allergic.

'A lot of people find when there are sunny, warm days and then a shower, the next day they feel crook,' he said.

Source: © Marnie McKimmie, *The West Australian*, 17 September 2005

1. How could the investigation reported in the article be used to help answer the question, 'What is science?'?
2. What sorts of things would you need to know if you wished to repeat Professor Murray's observations in order to verify his results?
3. Professor Murray was involved in both observation by looking at pollen grains under a microscope, and experimentation by checking what happened when water was added to the pollen. In testing pollen to see what happens when water is added, what control would be needed if the results were to be valid?
4. Can you think of any practical applications for Professor Murray's discovery?
5. The results of research often raise new questions to be answered. Suggest two questions that now need to be solved as a result of Professor Murray's research.
6. Propose a hypothesis based on one of the questions you suggested in your answer to Question 5.
7. The journalist has written an article describing Professor Murray's results for the general public. What additional information should be included in a report intended for other scientists working on the links between pollen grains and asthma?

ACTIVITY 1.3 Designing controlled experiments

One of the first controlled experiments in science was performed in 1668 by Italian doctor Francesco Redi. In Redi's time, it was believed that living organisms arose from non-living matter, an idea known as spontaneous generation.

Redi put meat into a number of flasks. He sealed half of the flasks and left the other half open. He then repeated his experiment, but instead of sealing half the flasks he covered them with gauze so that air (but not flies) could enter. Redi found that maggots developed in the open flasks but not in the flasks that were sealed or covered with gauze.

1. Suggest the hypothesis that Redi was testing.
2. List the variables that Redi controlled in his experiments.
3. What other variables do you think Redi should have controlled?
4. What conclusion could Redi draw from his experiment?
5. Make a list of further questions to be answered that arise as a result of Redi's experiments.

The idea of spontaneous generation lingered in the belief that micro-organisms arose spontaneously in the medium in which they were found. It was not until the 1860s that Louis Pasteur finally quashed the idea of spontaneous generation. Pasteur's experiments showed that bacteria did not develop in a flask of nutrient solution if it was sterilised by boiling and if air entering the flask was filtered to remove bacteria.

6. What was Pasteur's independent variable?
7. What was Pasteur's dependent variable?
8. List the variables that Pasteur would have controlled so he could make a valid conclusion from his experiment.

9780170449090

ACTIVITY 1.4 Testing a hypothesis

A human biologist was testing the following hypothesis: 'A decrease in environmental temperature causes an increase in the level of hormone X in the blood.'

1 Suggest one prediction that can be made from this hypothesis.

To test the hypothesis, 12 adults were kept in a room (Room 1) at 22°C for 12 hours. The subjects were then transferred to a second room (Room 2), where they were kept for another 12 hours at 10°C. The group consisted of six men and six women, all the same age. They were fed an identical diet in Rooms 1 and 2. After the 12 hours in each room, the level of hormone X in each subject's blood was determined.

2 Why were six men and six women used for the experiment, instead of just one of each sex?
3 What was the experimental test?
4 What was the control test?
5 What was the independent variable?
6 What was the dependent variable?
7 What variables were controlled (according to the description of the experiment)?
8 Can you think of any other variables that should have been controlled? If so, explain why.
9 Do you think the experiment would have been a fair test?
10 What results would have supported the hypothesis?
11 What results would have disproved the hypothesis?

ACTIVITY 1.5 Testing the product claims for Hairnu

Hairnu is a product that claims to stimulate the growth of hair.

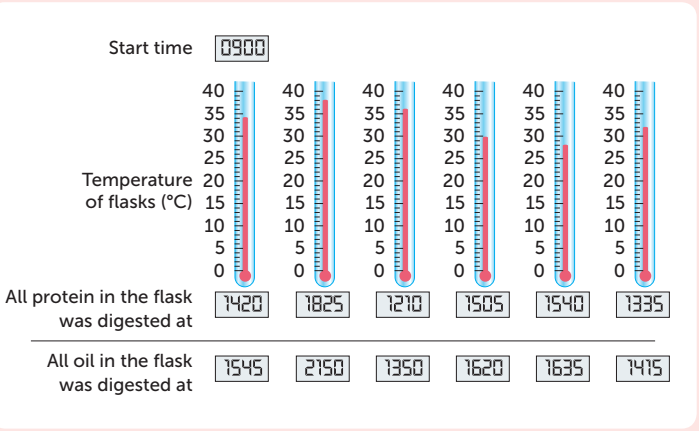
Design an experiment to test the claims that are made in this advertisement for Hairnu. In your design, make sure you cover all the following points.

- What will be your independent variable?
- What will be your dependent variable?
- State the hypothesis that you are testing.
- What variables will you need to control?
- How will you provide a control test so that you will be able to see whether Hairnu does what it claims to do?
- How will you measure your results?
- How many people will you need to test to get a reliable result?
- Draw a table to show how you would present your data.
- What results would support your hypothesis? What results would disprove your hypothesis?

ACTIVITY 1.6 Tabulating data

In this activity, you will practise drawing up a table to organise data.

Some students were investigating the effect of temperature on the activity of a digestive juice. They added 20 mL of the digestive juice to 50 mL of an emulsion containing standard quantities of oil and protein. The time taken for all the oil and all the protein to be digested was measured. This same procedure was repeated at a number of different temperatures. The results of the students' experiment are shown pictorially below.



1. What were the independent and dependent variables in the students' experiment?
2. Draw up a table to show the data they collected.

ACTIVITY 1.7 Graphing

1. The table below shows data on the weights of five babies from birth to one year.
   a. What is the dependent variable and what is the independent variable?
   b. Plot the data as a graph in the most appropriate manner.

BABY	WEIGHT (kg)				
	BIRTH	13 WEEKS	26 WEEKS	39 WEEKS	52 WEEKS
Amnah	2.1	5.9	8.5	9.6	10.7
Hamish	3.3	6.6	8.2	9.5	11.1
Chiu-Yin	3.4	6.3	7.9	9.3	11.2
Chloe	2.9	6.0	7.5	8.9	9.9
Ivy	3.2	6.7	8.4	9.8	11.6

2. Regular surveys of alcohol consumption are conducted in Australia. The annual consumption of wine, per person, over a number of years is shown in the table below.
   a. Identify the dependent and independent variables in these data.
   b. Plot the data as a graph in the most appropriate manner.

YEAR	1978	1981	1983	1986	1989	1993	1996	1999	2002	2004	2009
Litres	14.2	18.2	19.7	21.3	19.1	18.3	18.3	19.7	20.5	21.8	23.4

CHAPTER 1 SUMMARY

- Human biological science is the study of humans, both as individuals and populations, and their interactions with their environment.
- Science is both a process of inquiry and a body of knowledge.
- Our knowledge about humans is continuing to grow as we build on the discoveries of other scientists.
- There are different fields of human biology, such as anatomy, biochemistry, cytology, physiology and psychology.
- Scientific investigations involve literature review, observation, classification and/or experimentation.
- A hypothesis is a possible explanation for a problem that can be tested.
- The hypothesis can be used to predict the results.
- The variable is the factor that is tested, and the results are compared to the control experiment where nothing is changed.
- Controlled variables are factors that are kept the same in both the control and the test.
- The scientific method is a series of steps for scientific investigations. The steps are: recognition of a problem, collection of information, making a hypothesis, testing the hypothesis, collection of data, and analysis of data to reach a conclusion.
- Scientific investigations have an independent variable, which is changed and affects the dependent variable. All other variables are controlled. This ensures a fair test.
- Data collected can be quantitative (numerical) or qualitative (descriptive).
- Experiments should be repeated by conducting the same test a number of times. This means that any individual variation will have less effect on the results.
- Data can be organised in a table and represented in a graph. Line graphs are used for continuous data, column graphs are used for discrete data, and histograms are used for frequencies.
- The conclusion summarises what the data shows and whether it supports or disproves the hypothesis.
- The experiment is evaluated for its:
  - *validity*: whether it fairly tested the hypothesis or not
  - *accuracy*: whether the results are correct
  - *reliability*: whether the results for the same test are similar.
- Errors may be human errors, random errors or systematic errors.
- Any investigations involving humans or animals need to be ethically sound. They need to have voluntary participation, informed consent, minimal risk of harm, confidentiality and anonymity.
- Some investigations have a placebo, an inactive form of the test. In a blind experiment, the subjects do not know whether they are receiving the placebo or not. In a double-blind experiment, neither the researcher nor the subject knows who is receiving the placebo.

9780170449090

CHAPTER 1 GLOSSARY

Accuracy The extent to which the measurements are correct

Bar graph A graph for discrete data using horizontal bars

Blind experiment An experiment where the subjects do not know whether they are receiving the test treatment or the placebo

Classification The grouping of organisms based on the similarity of their characteristics; the placement of organisms into groups

Column graph A graph for discrete data using vertical bars

Conclusion A summary of how the data supports or disproves the hypothesis

Continuous data Quantitative data with an infinite number of possibilities

Control A procedure carried out to give a comparison in an experiment

Controlled variable A factor kept the same for both the control and the experimental groups in an experiment

Data Observations and measurements; the results of an experiment

Dependent variable In an experiment, the factor that changes in response to changes made to the independent variable; also called the responding variable

Double blind experiment An experiment where neither the subject nor the experimenter knows who receives the test treatment or the placebo

Ethical behaviour Behaviour that conforms to a set of moral principles or values

Ethics Moral principles or values

Fair test An experiment that only changes the independent variable and controls all other variables to test the hypothesis

Histogram A graph to represent the frequency distribution of data

Human biological science The scientific study of humans and their interaction with their environment

Human error An error due to the limitations of human ability

Hypothesis A possible explanation to account for observations; plural hypotheses

Independent variable In an experiment, the factor being investigated; the factor deliberately changed to determine its effect; also called the experimental variable or the manipulated variable

Interpretation An attempt to explain the observations

Line graph A graph used to represent continuous data

Observation The process of using the senses to acquire information

Placebo A substance or procedure that has no therapeutic effect but is used as a control test

Placebo effect A change or improvement in patients who are given a placebo or 'dummy' treatment

Prediction A guess at what might happen in the future

Qualitative data Observations that do not involve numbers or measurement

Quantitative data Data expressed in numbers; usually involves measurement

Random error An error in an experiment due to limits to the precision of the measurements

Reliability The extent to which an experiment gives the same result each time it is performed

Repetition Doing the same experiment many times

Scientific method A process of conducting valid investigations

Systematic error An error that occurs in an experiment because of the way the experiment was designed

Test A method used to collect data to determine whether a hypothesis is supported or not

Theory A hypothesis becomes a theory when there is overwhelming evidence in support of it

Validity The extent to which an experiment tests what it is supposed to test

Variable Any factor that may change during an experiment

9780170449090

CHAPTER 1 REVIEW QUESTIONS

Recall

1 a What is science?
  b Why is human biology a science?
2 a Describe a hypothesis.
  b Why do scientists make hypotheses?
3 a Define 'literature review'.
  b When would you carry out a literature review?
4 List the characteristics of a good hypothesis.
5 Describe the two types of experimental error, including how the effects of each type can be minimised.
6 List the ethical principles that must be satisfied in any research project.

Explain

7 Use an example to explain why classification is an important part of science.
8 Explain why an experiment must have a control.
9 Why is repetition important in scientific investigation?
10 The results of experiments are expressed as measurements whenever possible. Explain the reason for this.
11 Explain the difference between a hypothesis and a scientific theory.
12 a Explain the difference between the experimental group and the control group in an experiment.
  b What is the purpose of the control group?
13 A university study was conducted to investigate the effect of altitude on breathing rate. Use this example to:
  a explain the difference between the independent and dependent variables in an experiment
  b describe the controlled variables in an experiment.

Apply

14 a Explain the difference between the validity and the reliability of the results of an experiment.
  b How would you make sure that the results of an investigation are valid?
  c How would you make sure that the results of an experiment are reliable?
15 An American doctor, William Bean, studied the growth of his fingernails for 35 years. He filed a horizontal line on his thumbnail just above the cuticle (the strip of skin at the base of the nail). By recording how long it took the mark to reach the tip of the thumbnail he was able to calculate the growth rate. He was eventually able to conclude:

> A 35-year observation of the growth of my nails indicates the slowing of growth with increasing age. The average daily growth of the left thumbnail, for instance, has varied from 0.123 mm a day during the first part of the study when I was 32 years of age to 0.095 mm a day at the age of 67.

Source: W Bean, 'Nail growth: Thirty-five years of observation', *The Guardian*, 24 February 2004

  a Suggest a hypothesis that Dr Bean was testing.
  b State the independent and dependent variables.
  c List some of the variables that should have been controlled in Dr Bean's study.
  d Describe one source of random error in the investigation.
  e Measure the length of your thumbnail. Assume that your thumbnail grows at the same rate as that of the 32-year-old Dr Bean. How long did it take the tip of your thumbnail to grow from the cuticle to its present position?

9780170449090

f Do you think your fingernails and toenails grow at the same rate? Propose a hypothesis and outline an investigation that you could do to test your hypothesis.

16 Suppose you wished to find out whether people could tell the difference between normal instant coffee and decaffeinated instant coffee.
   a Propose a hypothesis and outline a blind experiment that you could do to test your hypothesis.
   b How could you make your experiment into a double-blind experiment?

17 Some scientists were testing a new drug called Presslo. It was hoped that Presslo would reduce blood pressure in people whose blood pressure was too high. The scientists selected two groups of people, all of whom were quite healthy but had high blood pressure. All the people were aged between 50 and 55 years. There were 100 people in each group and each group had equal numbers of males and females. One group was given a Presslo tablet at 8 a.m. each day. The control group was given a sugar pill at 8 a.m. each day. The blood pressure of the people in both groups was measured and recorded at the same time each day.
   a What was the independent variable in this experiment?
   b What was the dependent variable in the experiment?
   c List four variables that were controlled in the experiment.
   d List two variables that were not controlled in the experiment.
   e What was the purpose of the control group?
   f Why did the scientists have so many people in each group?

Extend

18 Why are reports of scientific investigations published?

19 Why do scientists use such a lot of technical terms?

20 Is history a science? Is music a science? Give reasons for your answers.

21 The word ‘malaria’ comes from two Italian words: male meaning ‘bad’, and aria meaning ‘air’. The ancient Greeks and Romans believed that malaria was caused by ‘bad air’ associated with swamps and marshes. We now know that this is not the case.
   a Find out how the Ancient Greeks and Romans tried to stop the spread of malaria.
   b Use resources to find out what causes malaria.
   c What were some of the experiments that were done to determine the cause of malaria?
   d Which scientists were instrumental in discovering the cause of malaria? How was their discovery communicated to others?

9780170449090

2 CELLS MAKE UP THE HUMAN BODY

UNIT 1 CONTENT

SCIENCE INQUIRY SKILLS

- conduct investigations, including monitoring body functions; use microscopy techniques; and perform real or virtual dissection, safely, competently and methodically for the collection of valid and reliable data
- select, construct and use appropriate representations, including labelled diagrams and images of various cells, tissues and organ systems, to communicate conceptual understanding, solve problems and make predictions
- communicate to specific audiences, and for specific purposes, using appropriate language, nomenclature, genres and modes, including scientific reports

SCIENCE UNDERSTANDING

Cells and tissues

- the human body is comprised of cells, tissues and organs within complex systems that work together to maintain life
- cell organelles maintain life processes and require the input of materials and the removal of wastes to support efficient functioning of the cell
- the cell membrane separates the cell from its surroundings with a structure, described by the fluid mosaic model, which allows for the movement of materials into and out of the cell by diffusion, facilitated diffusion, osmosis, active transport and vesicular transport (endocytosis/exocytosis)
- factors affecting the exchange of materials across the cell membrane include surface area to volume ratio, concentration gradients, and the physical and chemical nature of the materials being exchanged
- the various tissues of the human body perform specific functions and can be categorised into four basic tissue types: epithelial, connective, muscular and nervous

Source: School Curriculum and Standards Authority, Government of Western Australia

9780170449090

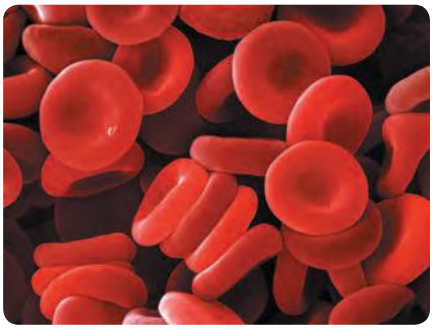
2.1 CELLS

A cell is the smallest structure that can perform life's functions. Some living organisms are made up of only one cell, while others are made up of many cells that work together. This means that cells are the basic structural and functional units of all living organisms, including plants and animals. This is a basic principle of biology and is known as the **cell theory**.

Key concept

The cell theory states that all organisms are made of cells, which are the basic unit of life and arise from pre-existing cells.

The human body contains countless millions of cells. In just 1 mL of human blood there are about 5 million red blood cells. There are up to 6000 mL of blood in an adult human, and blood is just one of the many tissues that make up the human body!



Science Photo Library/Power and Syred

FIGURE 2.1 Red blood cells are one type of cell

The structure of the human body, and the way in which it functions, result from the activities of all its cells. Everything we do results from the combined and coordinated actions of our cells. Each cell, however, is an individual unit with requirements that must be satisfied if it is to function normally.

All cells are very small – so small that you need a microscope to see most of them. They vary in size and shape. Despite these variations, all human cells have a similar basic structure.

Questions 2.1

RECALL KNOWLEDGE

1 State the cell theory.
2 Why do we use a microscope to view cells?

APPLY KNOWLEDGE

3 Compare and contrast cells in a human body with bricks used to build a house.

2.2 CELL STRUCTURE

Cell theory
Watch this short video about cell theory.

The structure of cells allows them to meet the requirements of life.

Cells are made up of the following parts:

- *cell membrane* – the outer boundary of the cell
- *cytoplasm* – those parts of the cell within the cell membrane, except for the nucleus; includes the jelly-like fluid and the organelles suspended in it
- *organelles* – structures suspended in the cytoplasm that carry out particular functions
- *cytosol* – the liquid part of the cytoplasm
- *cytoskeleton* – internal scaffolding of protein fibres within the cytoplasm
- *inclusions* – chemical substances occurring as granules or liquid droplets in the cytoplasm.

Figure 2.2 shows all the parts of a cell and summarises their structures and functions. It is important to realise that the diagram is a *model* of cell structure – no cell would be exactly like the one shown. Cells have many different shapes and differing numbers of the various organelles.

9780170449090

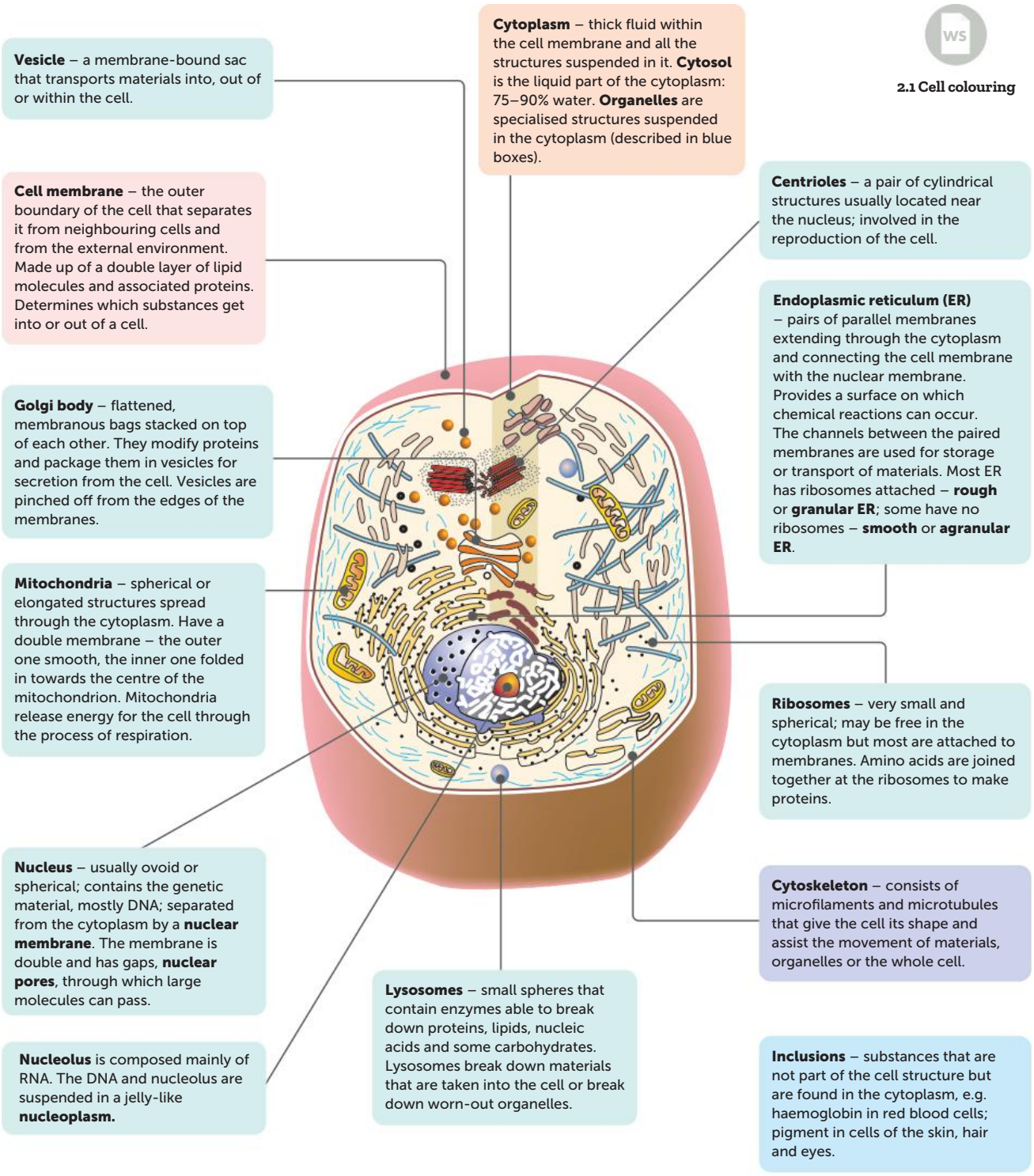


FIGURE 2.2 Model of cell structure and functions

Cell membrane

The **cell membrane**, or **plasma membrane**, separates the cell contents from the environment outside the cell and from neighbouring cells. It encloses the contents of the cell and controls what is able to enter and leave. The cell membrane is very thin – too thin to be seen clearly with a light microscope. The structure of the membrane will be discussed in detail later in the chapter.

9780170449090

Cells of most organisms have complex systems of internal membranes in addition to the plasma membrane. These internal membranes form structures inside the cell called organelles.

Cytoplasm

Cytoplasm is the jelly-like or watery material inside the cell that fills all the space between the nucleus and the cell membrane. It is made up of the cytosol and organelles.

Cytosol

The **cytosol** is the liquid part of the cytoplasm. It is 75% to 90% water, with a complex mixture of dissolved substances such as salts and carbohydrates. Other compounds, such as proteins and fats, do not dissolve but are suspended in the watery fluid.

The cytosol is where most of the metabolic reactions occur. It also plays a role in controlling the osmotic pressure of the cell and the flow of chemicals into and out of the cell.

Organelles

The structures within a cell are called **organelles**. Different types of organelle are specialised for particular functions. Many of the organelles are formed by the cell's internal membranes.

Nucleus

Almost all cells contain a single **nucleus**, although some, such as liver cells, have two or more nuclei and mature red blood cells have no nucleus at all. The nucleus is the largest organelle in the cell and is usually oval or spherical in shape. A **nuclear membrane** separates the nucleus from the cytoplasm. The nuclear membrane is actually a double membrane – two membranes separated by a space. Numerous gaps, or **nuclear pores**, in the nuclear membrane allow large molecules, such as messenger RNA, to enter and leave the nucleus.

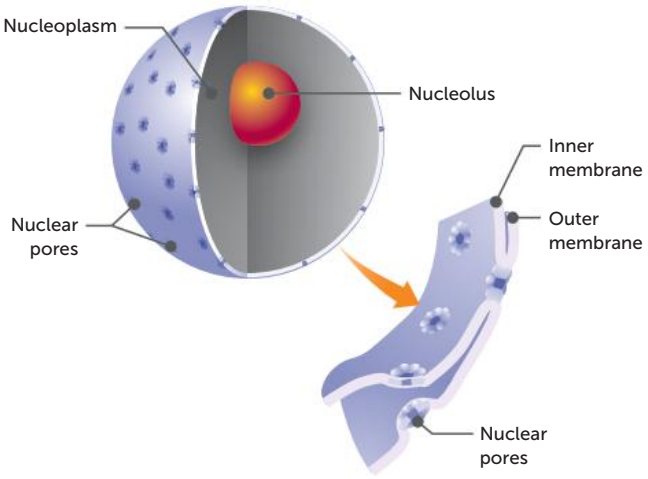


FIGURE 2.3 Structure of the nucleus showing the double membrane

Inside the nucleus is the **DNA** (**deoxyribonucleic acid**), which contains inherited information. When the cell is not dividing, the DNA is in the form of long threads called chromatin. In a dividing cell, the threads thicken and coil to form **chromosomes**. DNA contains the information that determines the type of proteins a cell can make. In this way the nucleus, with its DNA, controls the structure of the cell and the way it functions.

Inside the nucleus, the **nucleolus** plays a part in manufacturing proteins.

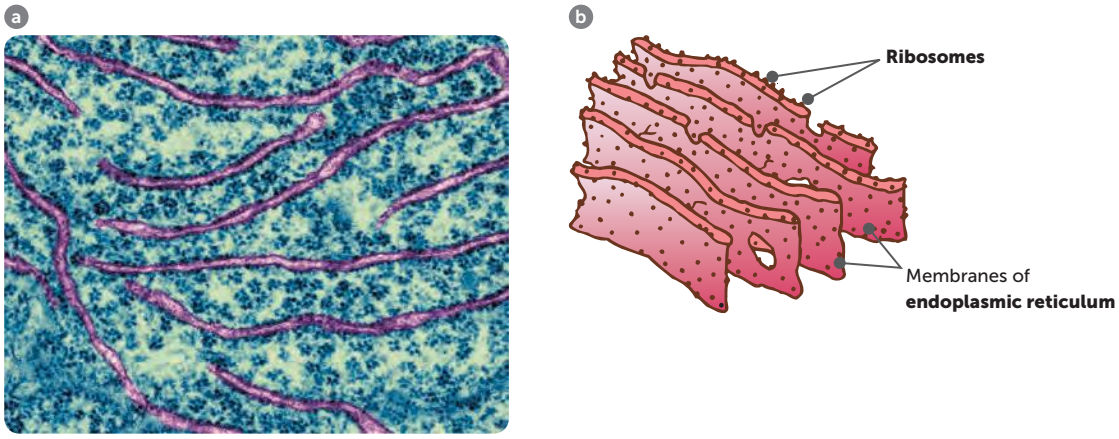
Ribosomes

Ribosomes are very small, spherical organelles. At the ribosomes, amino acids are joined together to make proteins. Ribosomes may be either free in the cytoplasm or attached to membranes within the cells such as the endoplasmic reticulum.

Endoplasmic reticulum

Pairs of parallel membranes extend through the cytoplasm of the cell from the cell membrane to the nuclear membrane. The network of channels formed by the parallel membranes is called the **endoplasmic reticulum**, or ER. It is thought that the membranes of the endoplasmic reticulum provide a surface for chemical reactions, while the channels are for storing or transporting molecules.

The endoplasmic reticulum can be classified as **rough endoplasmic reticulum**, when ribosomes are attached to the outside of some membranes, or **smooth endoplasmic reticulum**, when there are no ribosomes attached to the outside.

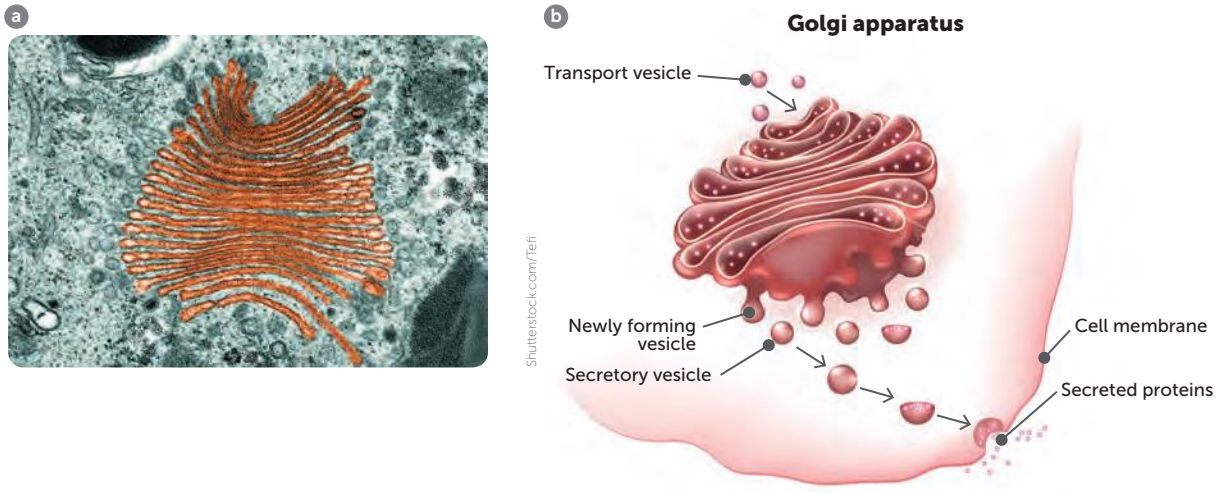


Science Photo Library/Dennis Kunkel Microscopy

FIGURE 2.4 **a** Electron micrograph showing endoplasmic reticulum with ribosomes attached; **b** Diagram showing the three-dimensional arrangement of the membranes

Golgi body

The **Golgi body** (sometimes called the Golgi apparatus) is a series of flattened membranes stacked one upon the other. Usually the Golgi body is positioned near the nucleus. Its function is to modify proteins and to package them for secretion from the cell. Proteins produced at the ribosomes pass through the channels of the endoplasmic reticulum to the Golgi body. At the edges of the membranes of the Golgi body, small sacs of liquid containing proteins are formed. These sacs are surrounded by a membrane and are called **vesicles**.



Science Photo Library

Shutterstock.com/Tefi

FIGURE 2.5 **a** Transmission electron micrograph showing section through a Golgi body; **b** Diagram indicating the three-dimensional shape of the Golgi body

Lysosomes

Lysosomes are small spheres, bound by a membrane, that are formed from the Golgi body. They contain digestive enzymes that are able to break down large molecules. When particles, or liquids, are taken into a cell they form vesicles in the cytoplasm. Lysosomes can join with these vesicles, and the digestive enzymes they contain break down the material inside the vesicle. Lysosomes also digest worn-out organelles in a similar way.

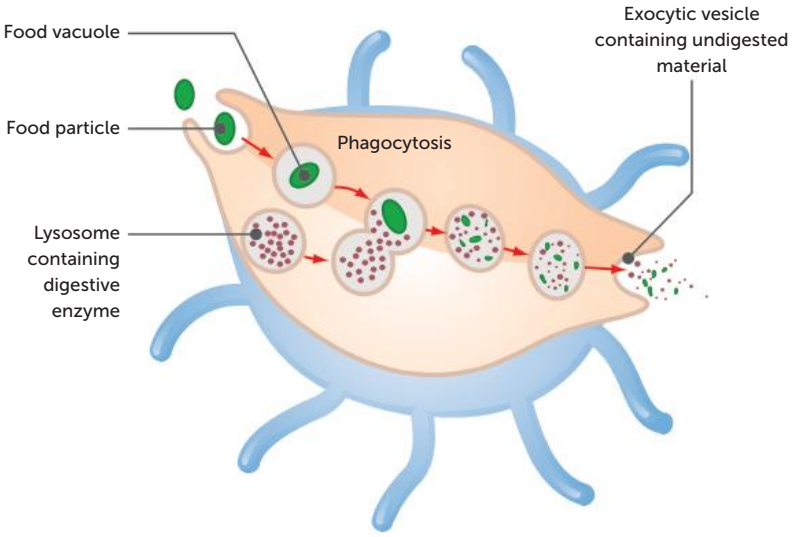
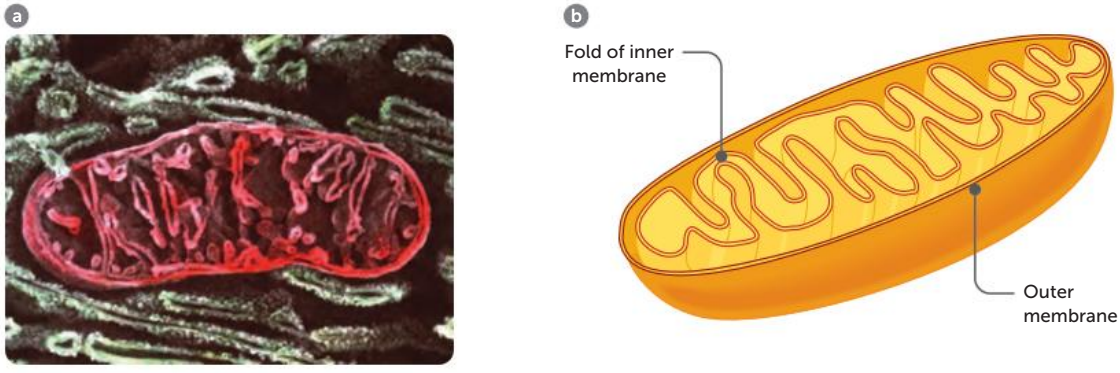


FIGURE 2.6 Digestive enzymes within lysosomes break down material inside vesicles

Mitochondria

Mitochondria (singular: mitochondrion) are spherical or sausage-shaped structures that are spread throughout the cytoplasm. Each has a double membrane. The smooth outer membrane surrounds the mitochondrion, while the inner membrane, called the cristae, is arranged into a series of folds that extend into the interior of the organelle.

Some of the chemical reactions of **cellular respiration** occur in the mitochondria. The folding of the inner membrane produces a large surface area on which these chemical reactions can take place. Because the reactions of the mitochondria make energy available for the cell's activities, these organelles are often called the 'powerhouses' of the cell.



Science Photo Library/Professors P Motta & T Naguro

FIGURE 2.7 **a** Electron micrograph showing section through a mitochondrion; **b** Diagram showing a three-dimensional view of a mitochondrion

Cilia and flagella

Some cells have fine projections that can beat back and forth to move either the whole cell or substances over the surface of the cell. If the projections are short and numerous, resembling tiny hairs, they are called **cilia**. If they are longer, and there is only one or two of them, they are called **flagella**. One place in which cilia occur is in the cells lining the trachea, where they move mucus and trapped particles towards the throat. In humans, only one type of cell – the sperm cell – has a flagellum; this enables the sperm to swim to the egg.

Organelles in a cell
Use this animation to review the organelles in a cell.

Cell structure
Use this website to review the structure of cells.

Cytoskeleton

The **cytoskeleton** is a framework of protein fibres that gives the cell its shape and assists cell movement. It consists of:

- **microtubules** – hollow rods that keep organelles in place or move them around the cell
- **microfilaments** – which move materials around the cytoplasm or move the whole cell.

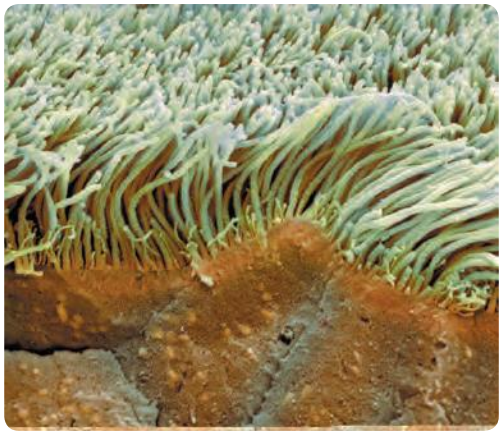

Alamy Stock Photo/Science Photo Library

FIGURE 2.8 Scanning electron micrograph of the wall of the trachea showing the cilia

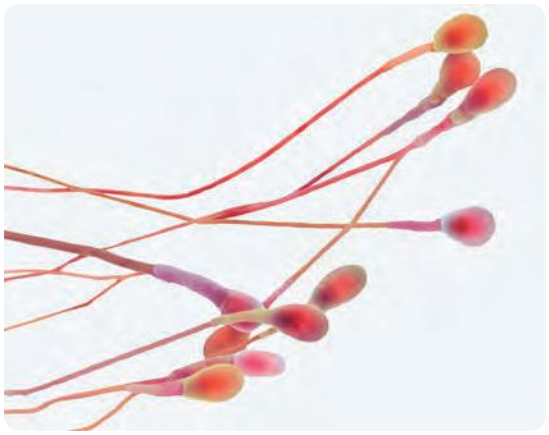

Science Photo Library/Dennis Kunkel Microscopy

FIGURE 2.9 Scanning electron micrograph showing sperm cells with flagella

Inclusions

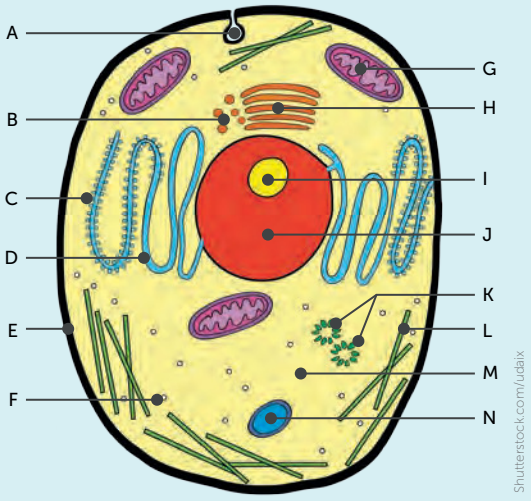
Inclusions are chemical substances that are not part of the cell structure but are found in the cytoplasm of the cell. Examples of inclusions include haemoglobin, the red pigment in red blood cells, and the pigment melanin in cells of the skin, hair and iris of the eye.

Key concept

The structures of the cell work together to meet the cell's needs and allow it to fulfil its function.

Questions 2.2

RECALL KNOWLEDGE



Shutterstock.com/udaix

1 Label the structures on the diagram of a cell.
2 Describe the function of the:
   a nucleus
   b mitochondria
   c microtubules
   d cytoplasm.
3 Explain the difference between cilia and flagella.
4 State the difference between rough and smooth endoplasmic reticulum.
5 Describe the role of nuclear pores.

Activity 2.1
Observing cells

Activity 2.2
Making a model of a cell

APPLY KNOWLEDGE

6 Explain how ribosomes, the endoplasmic reticulum and the Golgi body work together.
7 Explain why muscle cells have a large number of mitochondria.
8 Explain why cells vary in their size and shape.

2.3 CELL REQUIREMENTS

For normal functioning, cells in the human body need to be in a stable environment that continually supplies the materials they need and continually removes any materials they produce.

The immediate environment of a cell is the fluid that surrounds it; the **tissue fluid** or **extracellular fluid**. Even cells that appear to be very close together when observed under a microscope have a thin layer of fluid between them. This fluid allows a continual exchange of materials into and out of cells.

Body systems work together to ensure that the cellular environment is kept constant. This is called **homeostasis**. The cells are maintained at a constant temperature, surrounded by fluids with a constant concentration.

To carry out their functions, cells need to take in certain substances from the tissue fluid. As they process these substances, they produce materials that must then be removed from the cell. Depending on their particular role, different cells have different requirements and produce different materials. However, there are certain substances that all cells require and all cells produce.

During cellular respiration, glucose and oxygen are used to produce carbon dioxide, water and energy. Therefore, cells need to be supplied with oxygen and glucose, while carbon dioxide and water are removed.

Many cells also produce substances that will be used elsewhere in the body, such as hormones and enzymes. Other wastes, in addition to carbon dioxide, are also produced. All these products are released into the tissue fluid.

Structure and function of the cell membrane

Each cell is surrounded by a cell membrane that separates the internal and external environment. Substances that enter or leave the cell must pass through this membrane; therefore, it is very important in determining which substances will get into or out of a cell.

The cell membrane and all the membranes within the cell have a similar structure. Even with an electron microscope the detailed structure of cell membranes is too small to be seen. For this reason, models have been proposed to account for the behaviour and composition of the cell membrane. In science, a model is a simple explanation of a complex idea. The currently accepted model for cell membrane structure is called the **fluid mosaic model**. The membrane is said to be *fluid* because the molecules of which it is made are constantly changing position, and it is said to be *mosaic* because it is composed of many different kinds of molecules.

The main structure of the membrane is composed of **phospholipid** molecules, which are **lipid** molecules containing a phosphate group. The phospholipids are arranged in two layers, known as a **bilayer**. Each phospholipid molecule has a head that is **hydrophilic** (water-loving), and a tail that is **hydrophobic** (water-hating). The phospholipids are arranged in the two layers with their heads on the outside and tails on the inside. They drift from place to place with their heads and tails moving, keeping the membrane fluid.

Key concept

The fluid mosaic model explains the structure and function of the cell membrane.

Embedded in the phospholipid bilayer of the membrane are cholesterol and protein molecules. The cholesterol molecules are wedged between the phospholipids. These molecules are important for the function integrity and stability of the membrane. Cell membranes have a variety of protein molecules, including receptor proteins, channel proteins, carrier proteins and cell-identity markers. Some of these molecules extend from one side of the membrane to the other, while others are bound to the membrane surface. Only about 2% of the molecules in the membrane are proteins, yet they make up about 55% of the mass of the membrane. This is because proteins are very large molecules.

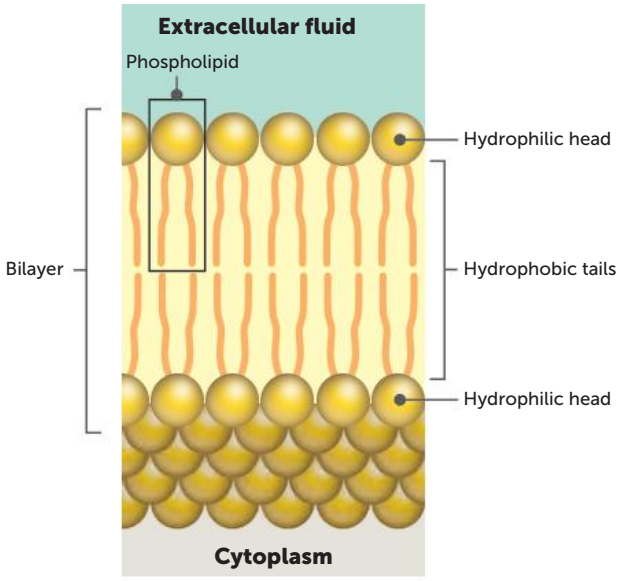


FIGURE 2.10 The phospholipid bilayer that makes up the basic structure of the cell membrane

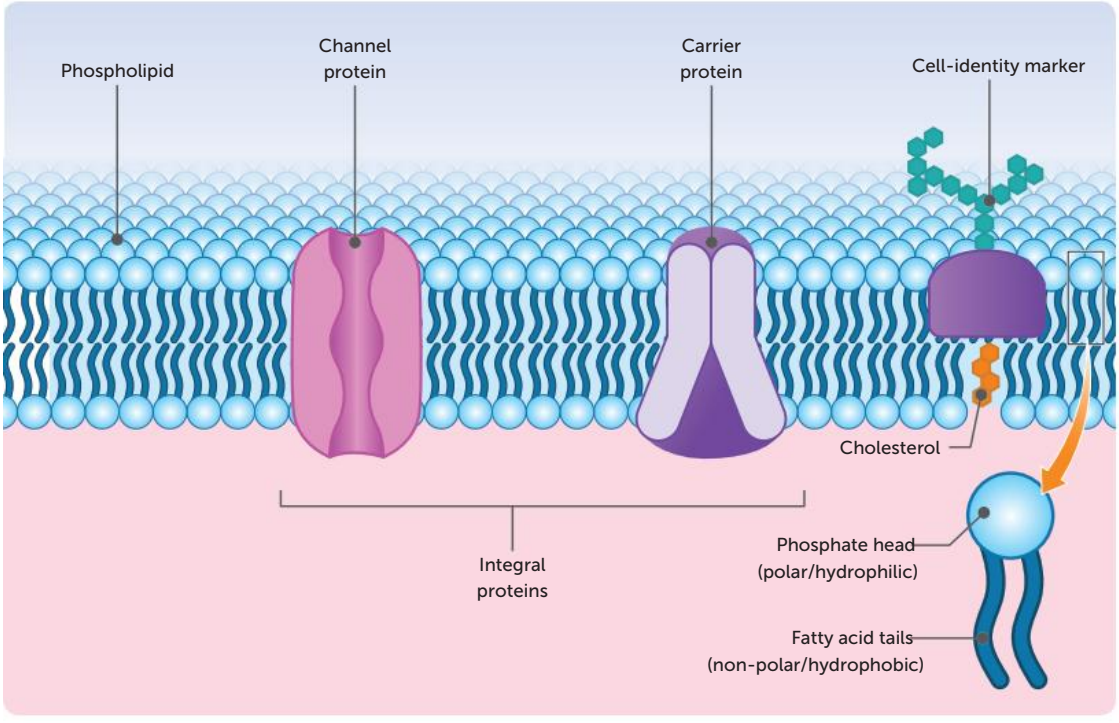


FIGURE 2.11 Model of a cell membrane, showing the phospholipid bilayer and protein and cholesterol molecules

Structure of the cell membrane
Use this website to review the structure of the cell membrane.

Activity 2.3
Making a model membrane

Functions of the cell membrane

The cell membrane has the following main functions:

- *It acts as a physical barrier.* The membrane separates the cell cytoplasm from the extracellular fluid around the cell. Isolation of the cytoplasm from the surrounding fluid is important because their compositions are very different.
- *It regulates the passage of materials.* The membrane controls the movement of materials into and out of the cell – for example, the entry of ions and nutrients, the removal of wastes and the release of secretions.

9780170449090

- *It is sensitive to changes.* The cell membrane is the first part of the cell affected by any changes in the extracellular fluid. It also has receptors that are sensitive to particular molecules in its immediate environment.
- *It helps support the cell.* The internal part of the cell membrane is attached to the microfilaments of the cell's cytoskeleton (see Figure 2.2 on page 27), thus giving support to the whole cell. There are also connections between the membranes of adjacent cells, providing support to the whole tissue.

Transport across the cell membrane

Cell membranes are described as being **differentially permeable,** semipermeable or selectively permeable. This means that they allow certain ions and molecules to pass through but restrict the movement of others.

Materials may pass through a cell membrane in different ways. Some transfer mechanisms are passive processes, while others are active. **Passive processes** do not use energy, whereas **active processes** use the cell's energy in the form of adenosine triphosphate (ATP).

Three basic processes result in transport of materials into or out of a cell:

- *Simple diffusion* – a passive process resulting from the random movement of ions and molecules; osmosis (also a passive process) is a special case of diffusion where water passes across the membrane.
- *Facilitated transport* – a process that requires special proteins in the cell membrane, either channel proteins or carrier proteins; it may be **passive transport** or **active transport**, depending on the exact nature of the mechanism.
- *Vesicular transport* – an active process in which materials are moved in membrane-bound sacs.

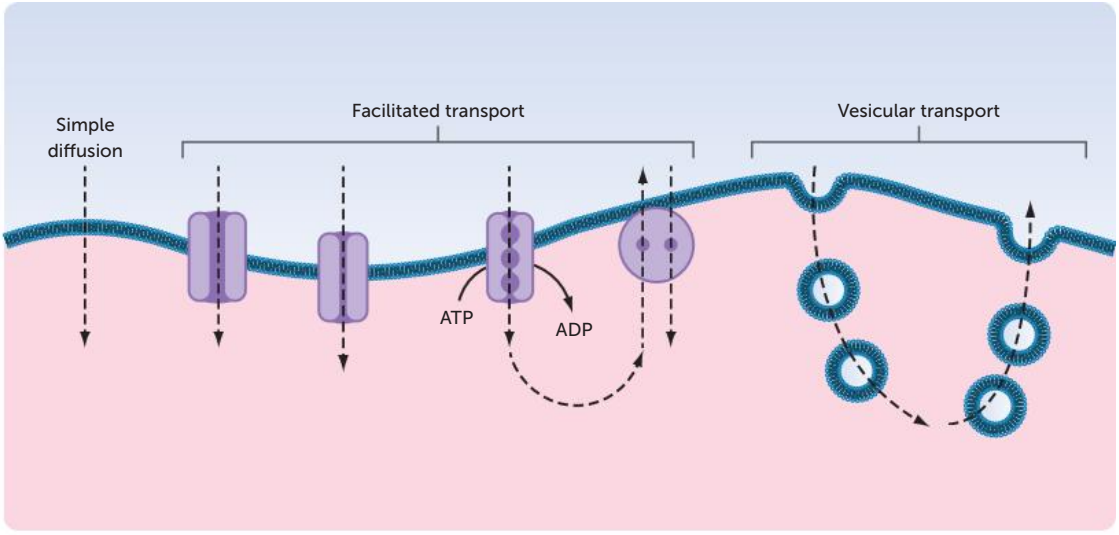


FIGURE 2.12
The different types of cellular transport

Key concept

The transport of materials into and out of cells is controlled; this may occur by diffusion, facilitated transport or vesicular transport.

Simple diffusion

Diffusion is the spreading out of particles so that they are evenly distributed over the space available. It occurs in gases and liquids because the molecules of gases and liquids are constantly moving. They move in random directions and in straight lines until they hit another molecule or the wall of the container. A deflected molecule then continues in a straight line until it hits another obstacle. Molecules moving away from an area in which they are concentrated experience fewer collisions than those moving towards the area of higher **concentration**. They therefore stay on their straight paths longer and move out into areas where the concentration of those molecules is lower. In this way, the molecules become evenly spread over the space available. The random movement of molecules continues, but the chances of collision are the same in whatever direction the molecule is travelling.

Figure 2.13 shows how a sugar cube dissolves in water and how the molecules of sugar spread out until they are evenly spread throughout the water. As the sugar dissolves, the sugar molecules near the cube are more concentrated than those near the surface of the water. The difference in concentration that brings about diffusion is called a **concentration gradient**, or **diffusion gradient** (Figure 2.14). The greater the difference in concentrations, the 'steeper' the diffusion gradient and the faster the rate of diffusion (Figure 2.15).

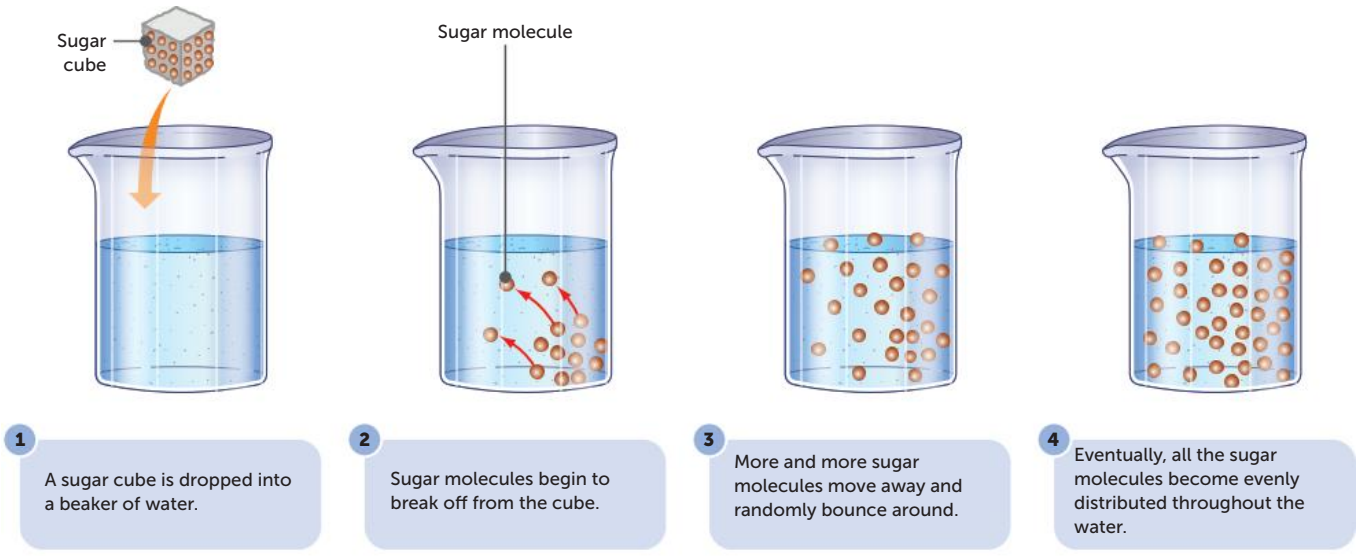


FIGURE 2.13 Diffusion results in the sugar being evenly distributed in the water

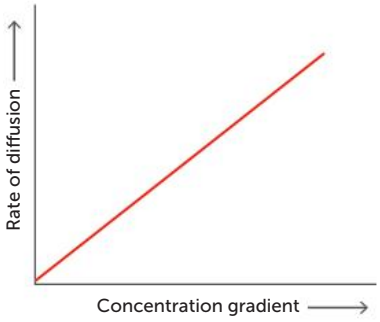


FIGURE 2.14 The higher the concentration gradient, the greater the rate of diffusion

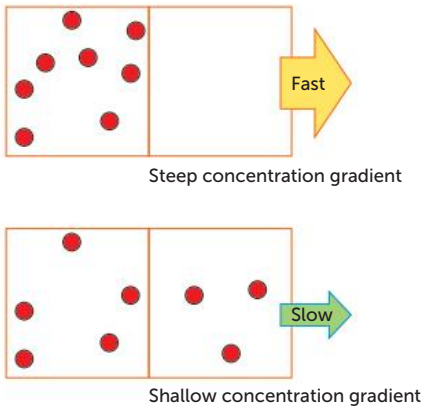


FIGURE 2.15 The difference in concentration between two areas determines the concentration gradient and, therefore, the rate of diffusion

9780170449090

The movement of liquid or gas molecules from places of higher concentration to places of lower concentration, along a concentration gradient, is more correctly called **net diffusion**. This is because there will also be some molecules moving against the concentration gradient in the opposite direction. While there is a difference in concentrations, there will be more particles moving from the area of high concentration to the area of low concentration. Once the concentrations are the same, the same number of particles will be moving in each direction.

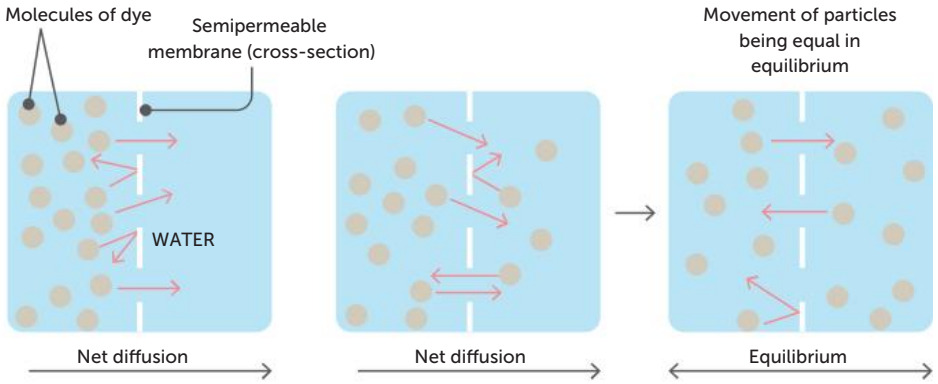


FIGURE 2.16 The movement of particles during diffusion and equilibrium

Alcohol, steroids and other fat-soluble substances can easily enter cells because they can diffuse through the lipid portions of the membrane. Oxygen and carbon dioxide can also diffuse through the phospholipid bilayer. This type of diffusion is referred to as **simple diffusion**.

- *Oxygen* diffuses into cells because it is continually used up inside the cell for respiration. The concentration of oxygen inside the cell is therefore lower than the oxygen concentration outside the cell. Because of this concentration difference, there is net diffusion of oxygen into the cell.
- *Carbon dioxide* is continually produced inside the cell by respiration. The higher concentration of carbon dioxide inside the cell means that there will be net diffusion of carbon dioxide out of the cell.

Water-soluble substances are unable to pass directly through the lipid portion of the membrane and hence require other modes of transport that are discussed in the next section.

Diffusion
Watch an animation of diffusion.

Osmosis
Watch an animation of osmosis.

Osmosis

Osmosis is a special type of diffusion: the diffusion of a **solvent** through a differentially permeable membrane in order to balance the concentration of another substance. As water is the most important solvent in the human body, osmosis can be considered to be the diffusion of water across a differentially permeable membrane. The water will move from an area where a solute such as sugar is in low concentration to an area where the solute is in high concentration. As more water moves into the high concentration, the solution will become diluted, lowering the concentration. At the same time, as the water moves out of the area of the low concentration the concentration will increase. This occurs because, if there are equal volumes in both areas, where there is more solute there will be less solvent. The concentration of water is therefore lower. Conversely, the area of the lower concentration of solute will have a higher concentration of water. In this way, water is moving from the area of high concentration to low concentration of water.

2.2 Transport across a membrane

Large polar molecules, such as glucose, and ions, such as sodium ions, are unable to cross the cell membrane directly as they are repelled by the hydrophobic tails in the phospholipid bilayer. However, water molecules are small enough to be able to pass through the cell membrane, since they can fit between the lipid tails. Water also crosses the membrane by passing through protein channels; this form of transport is discussed in the next section.

Figure 2.17 shows a beaker divided in two by a differentially permeable membrane. On one side of the membrane is pure water; on the other side is a sugar solution. Water molecules can

pass through the membrane, but the sugar molecules will stay on the same side of the membrane. Because of the difference in concentration, more water molecules will move from the water to the sugar solution than in the opposite direction. The sugar solution will gain water.

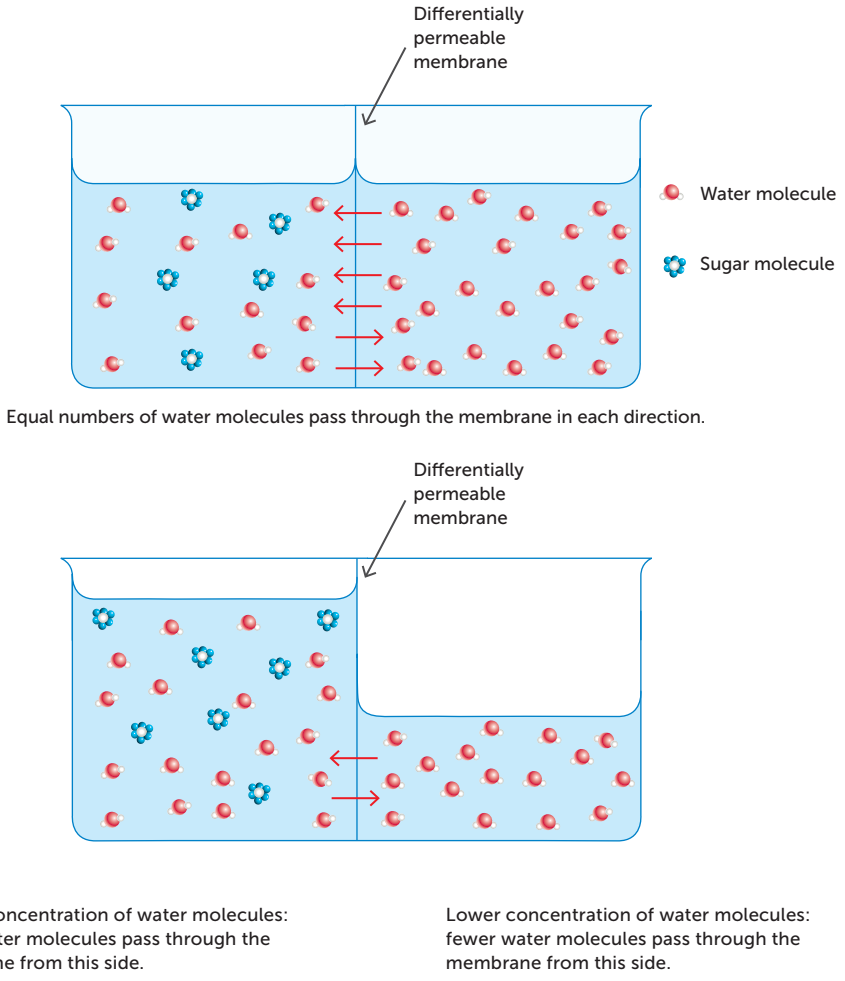


FIGURE 2.17 The process of osmosis

Note that in Figure 2.17, the level of liquid on the water side of the membrane has dropped, whereas the liquid level on the sugar side has risen. This higher level on one side of the membrane results from a pressure, known as **osmotic pressure**. The higher the concentration of solute (in this case, the sugar), the higher the osmotic pressure.

Facilitated transport

In **facilitated transport**, proteins in the cell membrane allow molecules to be transported across the membrane. These proteins are **channel proteins**, which form **protein channels**, and **carrier proteins**, which allow carrier-mediated transport.

Protein channels

To diffuse across a cell membrane, water-soluble molecules must pass through protein channels in the membrane, allowing **facilitated diffusion**. These channels provide a pathway for the hydrophilic particles to travel through to cross the cell membrane without coming in contact with the hydrophobic inner portion. The protein channels are very small in diameter, but water and ions can easily get through. Larger molecules are too big to fit through the channels.

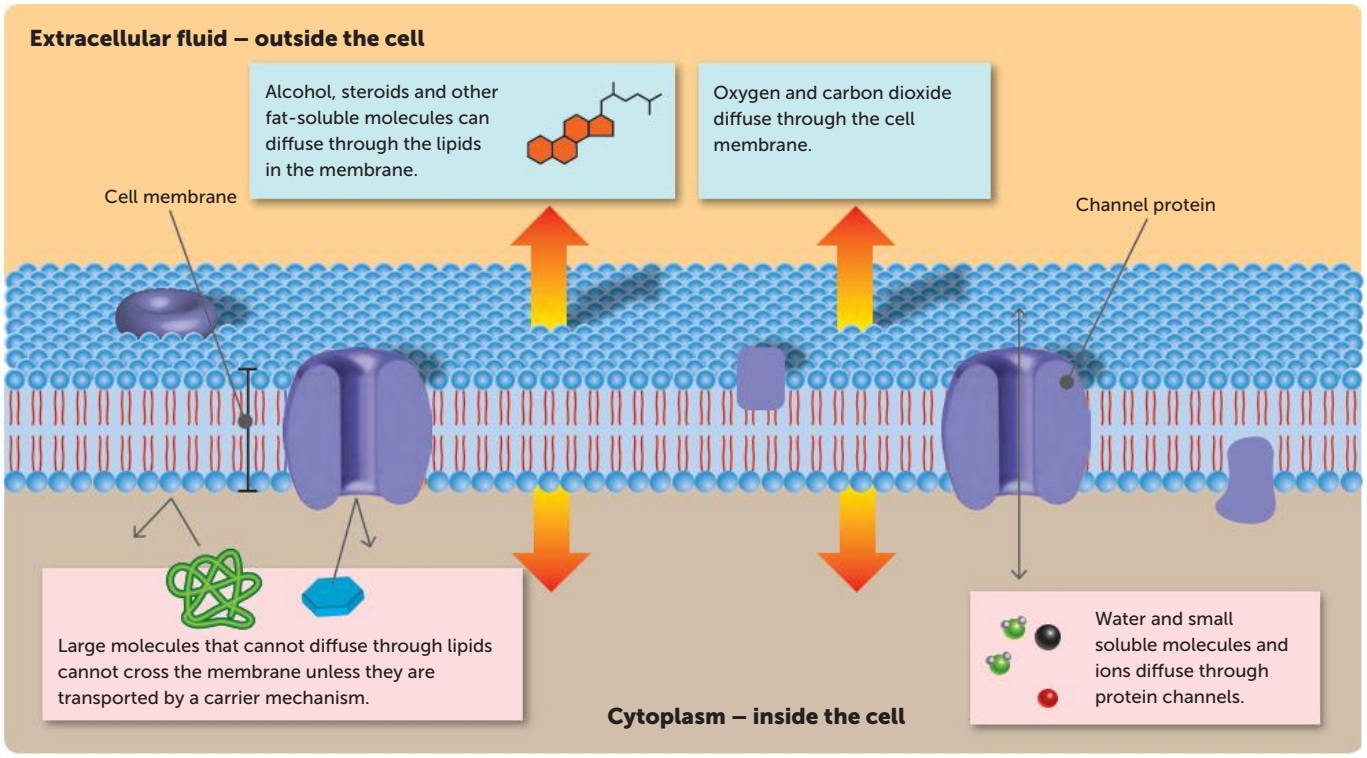


FIGURE 2.18 Diffusion of different molecules across a cell membrane

Activity 2.4
Investigating diffusion through a differentially permeable membrane

Carrier-mediated transport

While channel proteins provide a channel through the membrane, carrier proteins are only open on one side of the membrane at a time. When the specific substance binds to the binding site within the protein, the protein changes shape and opens to the other side. The substance can then be released on the side opposite to where is entered.

Some important characteristics of **carrier-mediated transport** are as follows:

- The carrier proteins are specific; they will only bind to a particular molecule. For example, the carrier that transports glucose cannot transport any other molecules, even simple sugars that are very similar to glucose.
- Carriers can become saturated. Once all the available carriers are occupied, any increase in the concentration of molecules to be transported cannot increase the rate of movement.
- Carrier activity is regulated by substances such as hormones. Hormones are important in coordinating the activities of carrier proteins.

There are two main types of carrier-mediated transport.

1. *Facilitated diffusion* occurs when substances are transported through a protein along the concentration gradient, from a higher concentration on one side of the membrane to a lower concentration on the other. This is a passive process, as it does not require the input of energy. During carrier-mediated facilitated diffusion, the molecule to be transported, such as glucose, attaches to a binding site on the specific carrier protein. The protein changes shape and the molecule is released on the other side of the membrane.
2. *Active transport* requires energy in the form of ATP because substances are transported across the membrane against the concentration gradient, from lower to higher concentration. The process of active transport is similar to that of facilitated diffusion via carrier proteins, but its big advantage is that it does not depend on a concentration gradient. Using active transport, a cell can take in or pass out substances regardless of their concentrations inside or outside the cell.

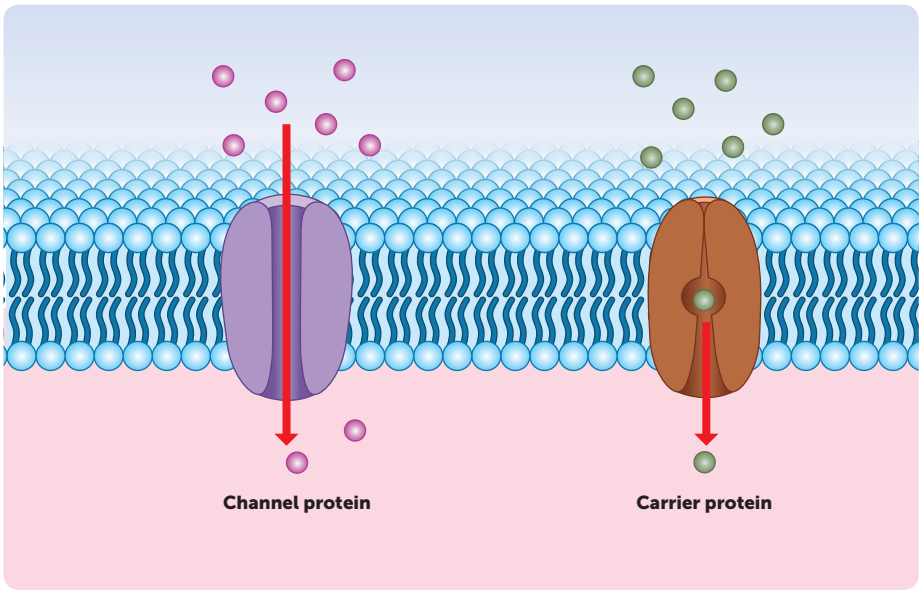


FIGURE 2.19 Comparison of channel proteins and carrier proteins

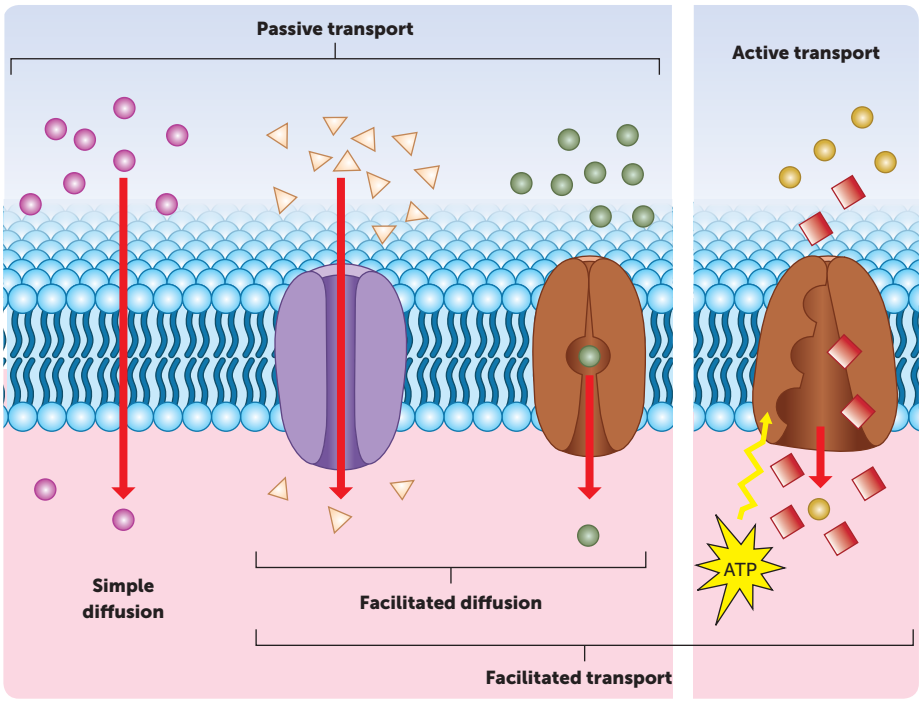


FIGURE 2.20 Simple diffusion and facilitated transport

Vesicular transport

Vesicular transport is the movement of substances across the cell membrane in membranous sacs called vesicles. This is an active process, because energy from the cell is needed to form the vesicles.

Endocytosis is taking liquid or solids into the cell by vesicular transport. The cell membrane folds around a droplet of liquid or a solid particle until the droplet or particle is completely enclosed. The vesicle formed then pinches off and is suspended in the cell's cytoplasm. Taking liquids into the cell in this way is called **pinocytosis**; when the vesicles contain solid particles it is called **phagocytosis**.

Exocytosis is when the contents of a vesicle inside the cell are passed to the outside. A vesicle that is formed inside the cell migrates to the cell membrane and fuses with the membrane. The contents of the vesicle are then pushed out into the extracellular fluid.

Transport across a cell membrane
Use this website to review the transport of materials across a cell membrane.

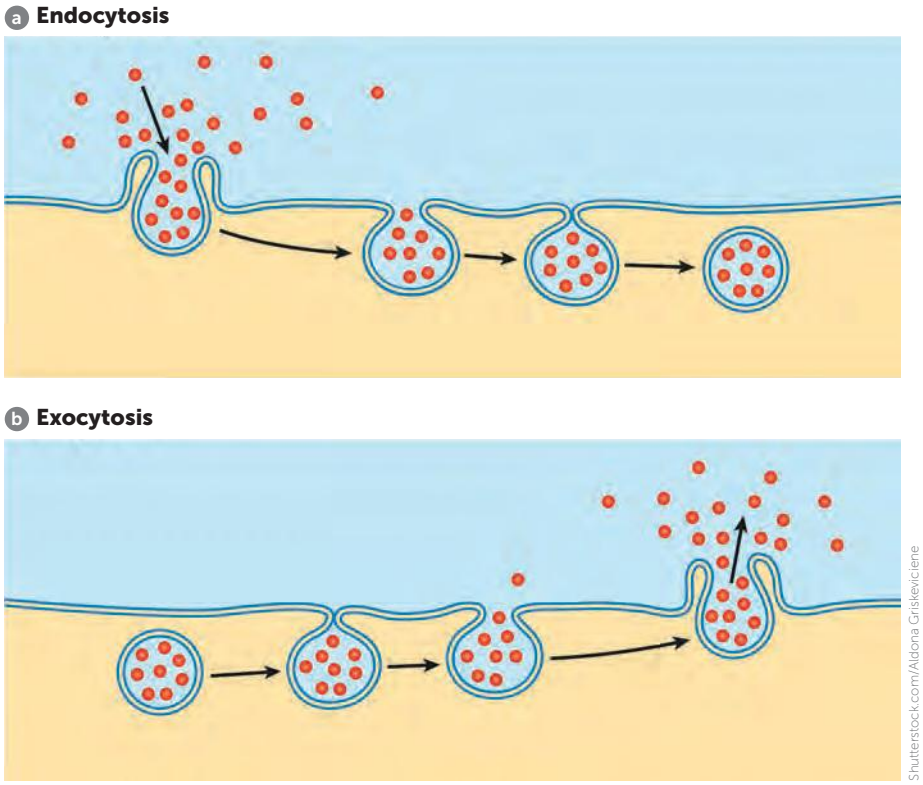


FIGURE 2.21
a Endocytosis;
b Exocytosis

TABLE 2.1 Summary of types of transport across the cell membrane

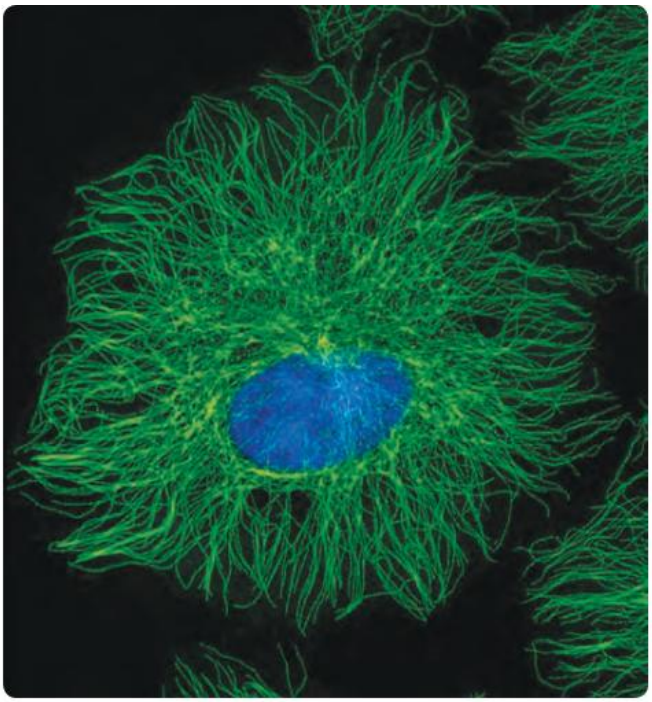
TYPE OF TRANSPORT	PASSIVE OR ACTIVE	SUBSTANCES TRANSPORTED
Simple diffusion		
Simple diffusion of solute	Passive	Water, oxygen, carbon dioxide, alcohol, fatty acids, steroids; ions such as sodium, potassium, calcium; lipids; soluble drugs
Osmosis	Passive	Water
Facilitated transport		
Facilitated diffusion	Passive	Glucose, amino acids
Active transport	Active	Certain ions, glucose, amino acids
Vesicular transport		
Endocytosis	Active	Cholesterol, iron ions; micro-organisms and cell debris but only by certain specialised cells
Exocytosis	Active	Secretions, such as mucus or digestive juices

Movement within the cell

Molecules and ions move within the cell mostly by diffusion. Remember that diffusion is the spreading of particles so that they are evenly distributed over the space available. Thus, as molecules of a substance are used up in one part of the cell, other molecules will spread to take their place. For example, as oxygen is used up by the mitochondria for respiration, a lower concentration of oxygen is created. Oxygen will then diffuse into the area of lower concentration from areas of higher concentration within the cell.

There are also structures that transport substances. The endoplasmic reticulum is used to transport substances within the cell – particularly proteins that the cell has made. These are transported to the Golgi body for secretion from the cell.

Microtubules are very fine tubes that help to maintain the shape of the cell and to hold the organelles in place. They also act like railway tracks, guiding organelles or molecules to particular places within the cell. Microtubules are not permanent structures but are able to be broken down or built up as needed in the various parts of the cell.



Science Photo Library/Thomas Deerinck, NCMIR

FIGURE 2.22 Fluorescent light micrograph of microtubules, showing the nucleus (blue) and microtubules (green)

Why are cells so small?

The cells in a human body vary greatly in size. Most human cells are extremely small; between 10 and 15 micrometres (μm) in diameter (1 μm is one-thousandth of a millimetre). Nerve cells may have extensions up to a metre long, and muscle cells up to 30 cm long. However, both nerve and muscle cells are too thin to be seen with the naked eye. Human egg cells have a diameter of up to 100 μm and may be just visible to the naked eye.

There is a limit to how big a cell can be. All the requirements and products of a cell must pass across the membrane that surrounds the cell. Thus, the relationship between the surface area of the cell and the volume is very important. Imagine that an apple is a cell. If the apple is cut in half, each piece has half of the original volume, but each piece has *more* than half of the original surface area. Cutting the apple in half has created extra surface area because of the two cut surfaces. If you continue to cut the apple into smaller and smaller pieces, the surface-area-to-volume ratio of the pieces gets bigger and bigger. In the same way, a small cell will have a larger surface-area-to-volume ratio than a large cell.

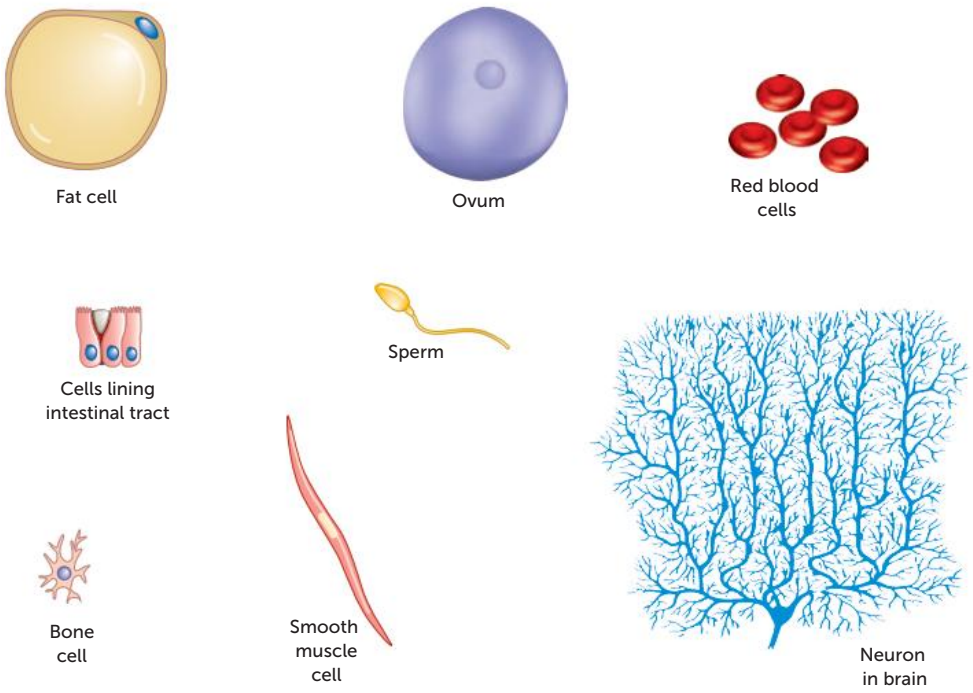


FIGURE 2.23 Human cells have varied sizes and shapes

Figure 2.24 illustrates how doubling the length of the side of a cube-shaped cell results in *eight* times the volume, but only *four* times the surface area. As a cell grows, its ability to exchange enough materials to support its increasing volume is diminished because the volume increases at a greater rate than the surface area. A large cell could not support itself because it would not have enough surface to absorb the nutrients required, and remove the wastes produced, for its large volume. To function effectively, most cells have to be microscopic.

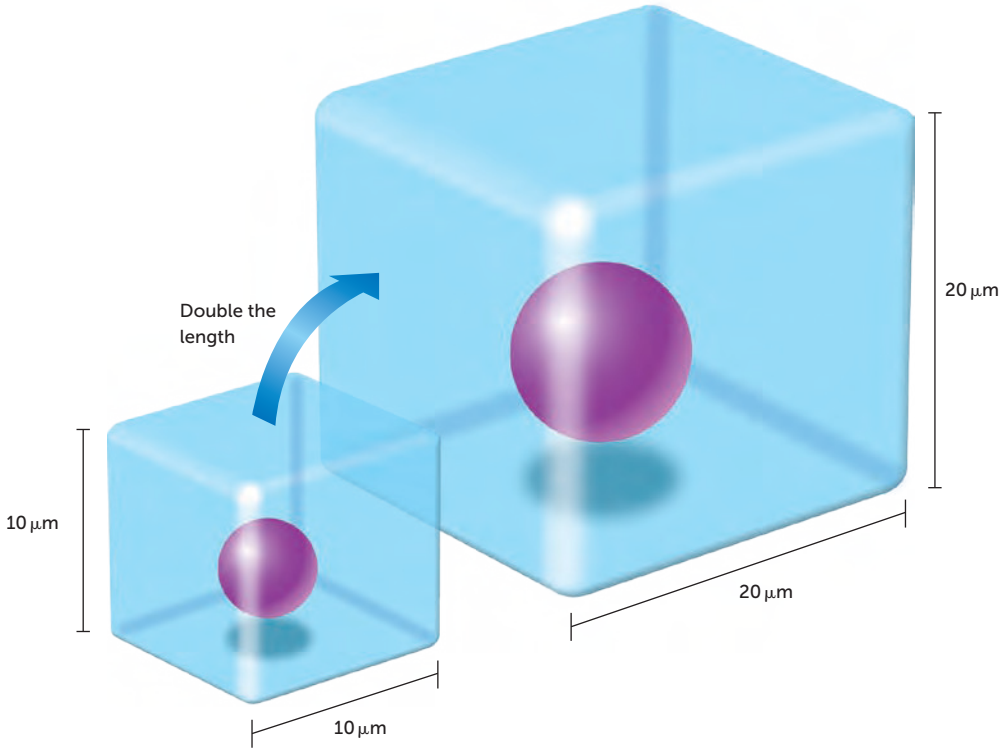


FIGURE 2.24 The relationship between the surface area of a cell and its volume. When the diameter of a cell is doubled, its volume is eight times greater, but its surface area is only four times greater

Activity 2.5
What size is it?

Activity 2.6
Investigating surface area and volume

Key concept

The surface-area-to-volume ratio limits the size of individual cells.

Questions 2.3

RECALL KNOWLEDGE

1. Define 'extracellular fluid'.
2. Draw a labelled diagram of the fluid mosaic model of the cell membrane.
3. List the substances that all cells need to take in.
4. State the functions of the cell membrane.
5. Which type of transport actively moves substances in membrane-bound sacs?

APPLY KNOWLEDGE

6. Compare and contrast the two types of carrier-mediated transport.
7. Explain why a cell will expand when placed in a solution of a low concentration of a solute such as sugar.
8. Explain why steroids are able to diffuse directly through the membrane, but glucose requires a carrier protein.

9780170449090

2.4 HOW CELLS MAKE A BODY

The body is organised on four structural levels.

1. *Cells*, the lowest structural level, are specialised to carry out different functions. Muscle cells are able to shorten in length; red blood cells are able to transport oxygen; cells of mucous membranes secrete mucus; and so on.
2. Cells with similar specialisations that carry out a common function are grouped together into *tissues*. For example, groups of muscle cells make up muscle tissue, groups of nerve cells make up nervous tissue, and groups of bone cells form bone.
3. Different types of tissues work together as *organs*. An organ is normally made up of two or more tissues. The stomach is an organ with epithelial tissue on the inside and muscular tissue in the wall; the heart is an organ made up of muscular tissue and nervous tissue.
4. The highest level of organisation is the *system*. A system is a group of organs that work together for a common purpose. For example, the respiratory system supplies oxygen and removes carbon dioxide from the blood. Some of the organs that make up the respiratory system are the lungs, diaphragm, intercostal muscles between the ribs, trachea, larynx and nose.

   The body systems are all integrated into the one living thing, the *organism*.

Tissues

A **tissue** is a group of cells that are similar in structure and that work together to carry out a particular task. The structure of the tissue and the function it performs can be used to classify it into one of four basic types. These four basic types of tissue are epithelial tissue, connective tissue, muscular tissue and nervous tissue.

Epithelial tissue

Epithelial tissue, or **epithelium**, is a covering or lining tissue. The outer layer of the skin is an epithelial tissue. Organs including the heart, kidneys, intestines, liver and lungs are covered with epithelium (see Figure 2.25). It also lines the inside of organs, so the inner layer of the heart, stomach, intestines and other hollow organs is made up of epithelium.

The cells that make up epithelium are very closely joined together. They vary in shape from thin and flat to column-shaped and cube-shaped, depending on the particular tissue. The cells that line the inside of your mouth are an example of thin, flat epithelial cells. Because they fit very closely together, they form a very smooth surface.

Shutterstock/Jose Luis Calvo

FIGURE 2.25 Example of epithelial tissue from the intestinal villus

Connective tissue

Connective tissue provides support for the body and helps to hold all the body parts together. One of the characteristics of connective tissue is that the cells are not close together like they are in epithelium. They are separated from each other by large amounts of material that is not made of cells. This non-cellular material is called **matrix**.

Connective tissues include bone, cartilage, tendons, ligaments and fat storage (adipose) tissue. Blood is often classified as a connective tissue (see Figure 2.27). The matrix of blood is the liquid in which blood cells are suspended.

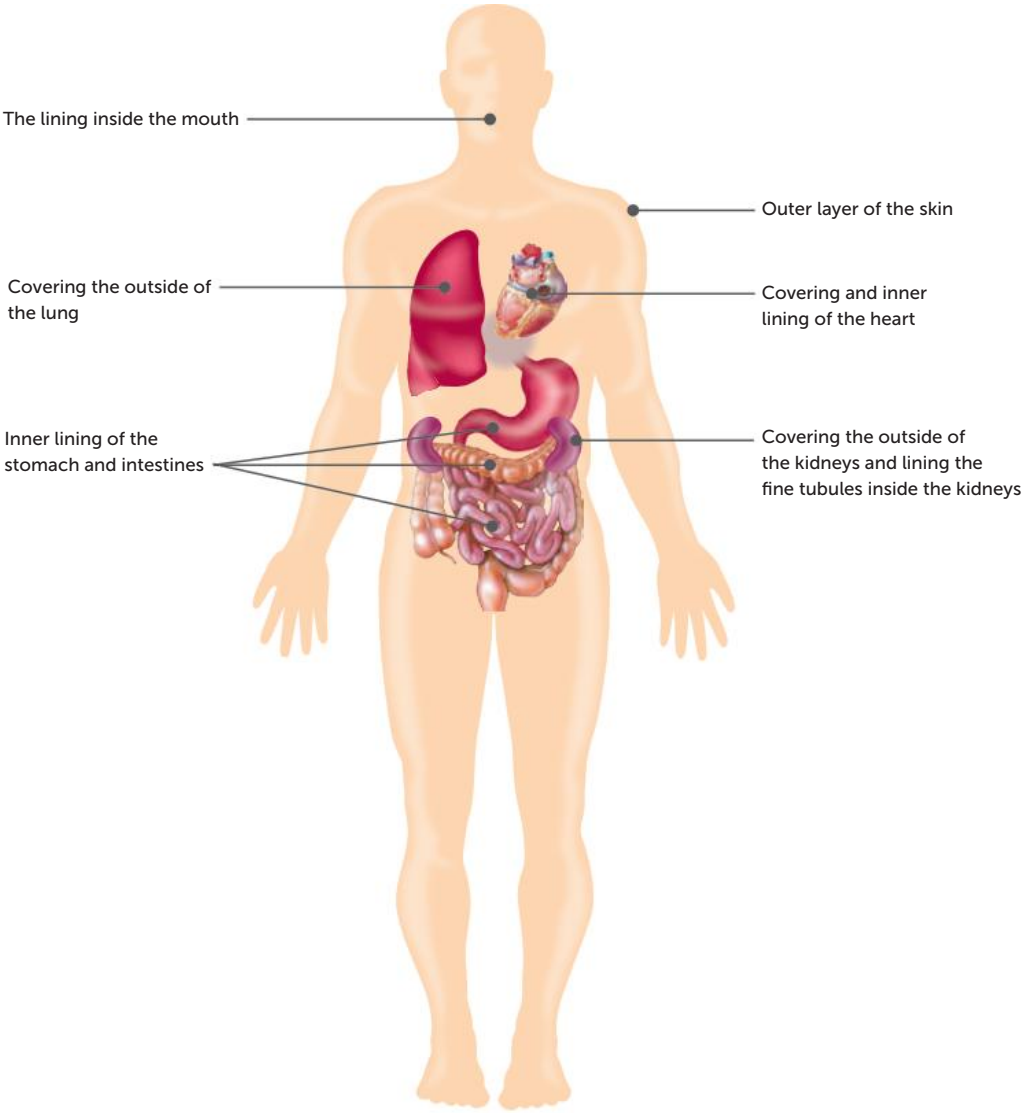


FIGURE 2.26 Location of some epithelial tissues

Muscular tissue

The cells of muscular tissue, often called **muscle fibres**, are long and thin and can contract to become shorter. There are three different types of muscular tissue: skeletal, smooth and cardiac muscle.

Skeletal muscle makes up the muscles that are attached to bones. These are the muscles that you can feel in your arms and legs. We have voluntary control over these muscles so that we can move parts of our bodies when necessary. Skeletal muscle is thus sometimes referred to as **voluntary muscle**. Under a microscope, skeletal muscle fibres are seen to have stripes, or striations, across them, so another name for this muscle is **striated muscle**. The nature of the striations will be discussed in Chapter 8.

Smooth muscle does not have any striations, and therefore, is also called **non-striated muscle**. It is found in the walls of the stomach and intestines, in the walls of blood vessels, in the iris of the eye, in the uterus and many other organs. We cannot contract smooth muscle voluntarily, and hence it may also be called **involuntary muscle**.

Cardiac muscle (also known as **heart muscle**) makes up most of the heart. When heart muscle contracts, it pumps the blood. Heart muscle cannot be voluntarily controlled.

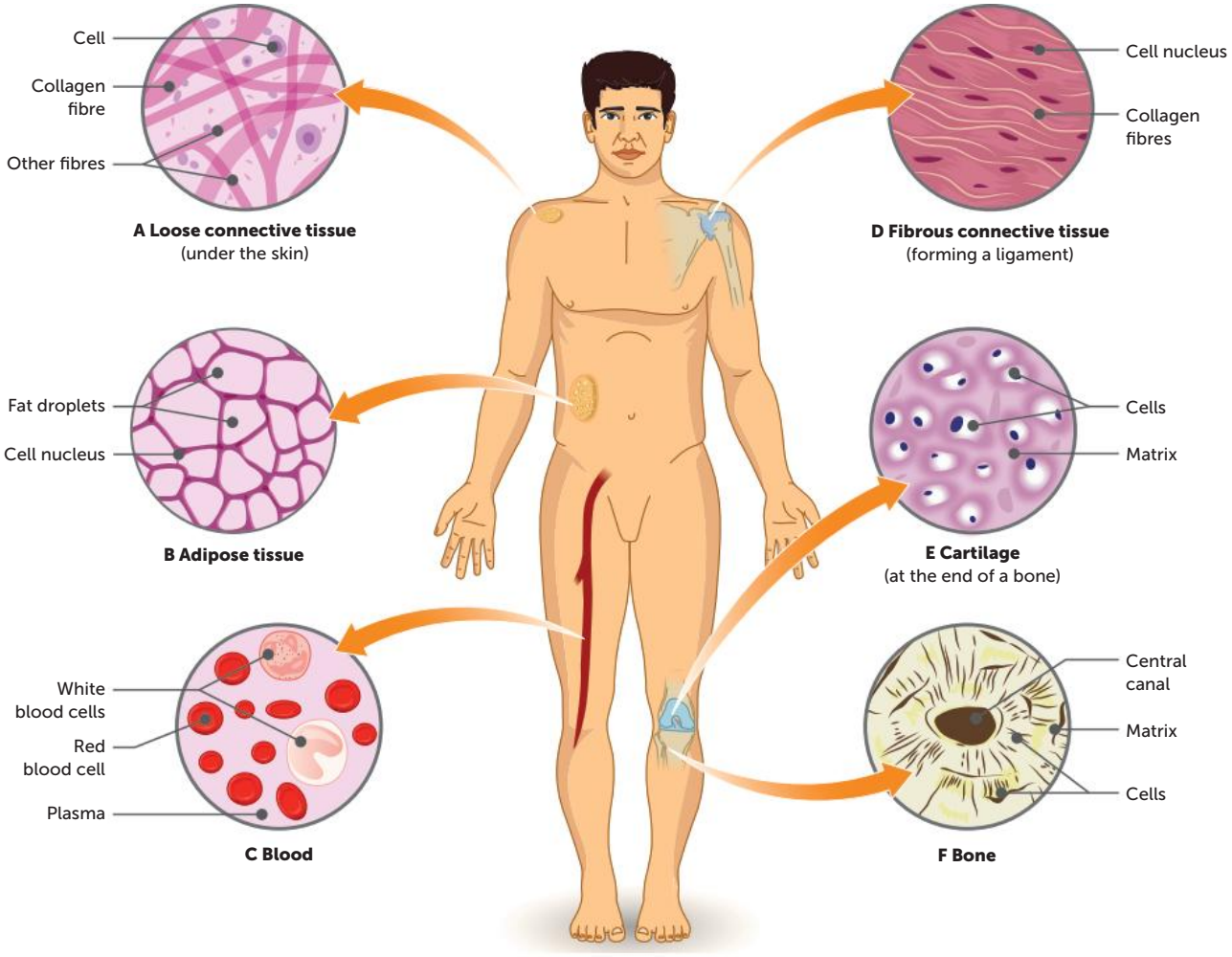
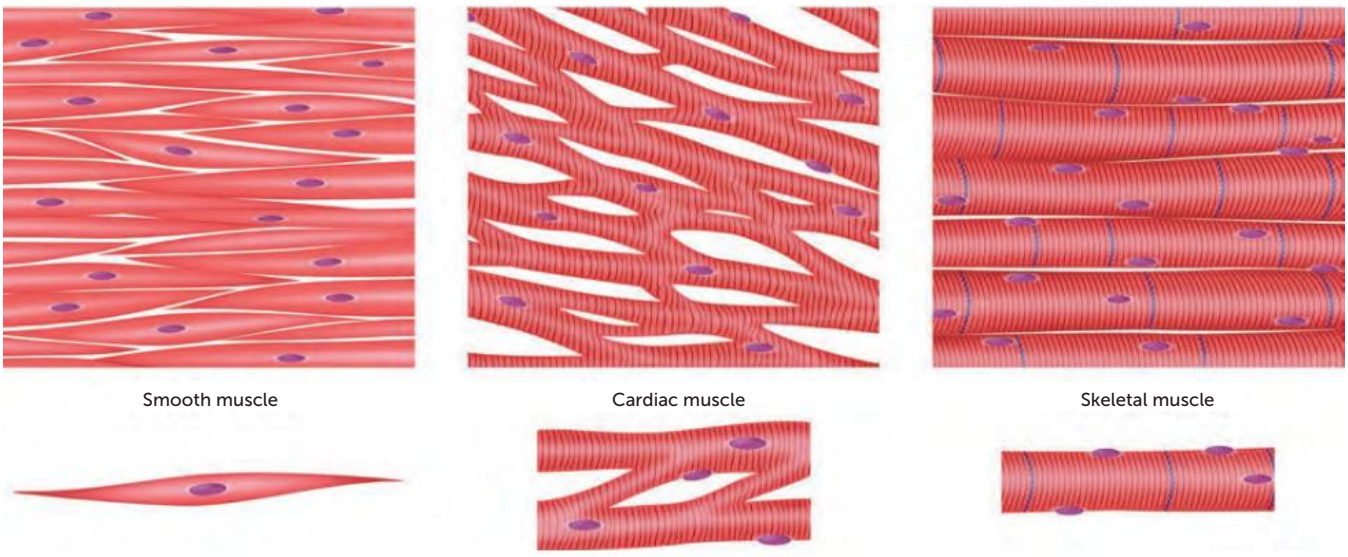


FIGURE 2.27 Examples of connective tissue in the body



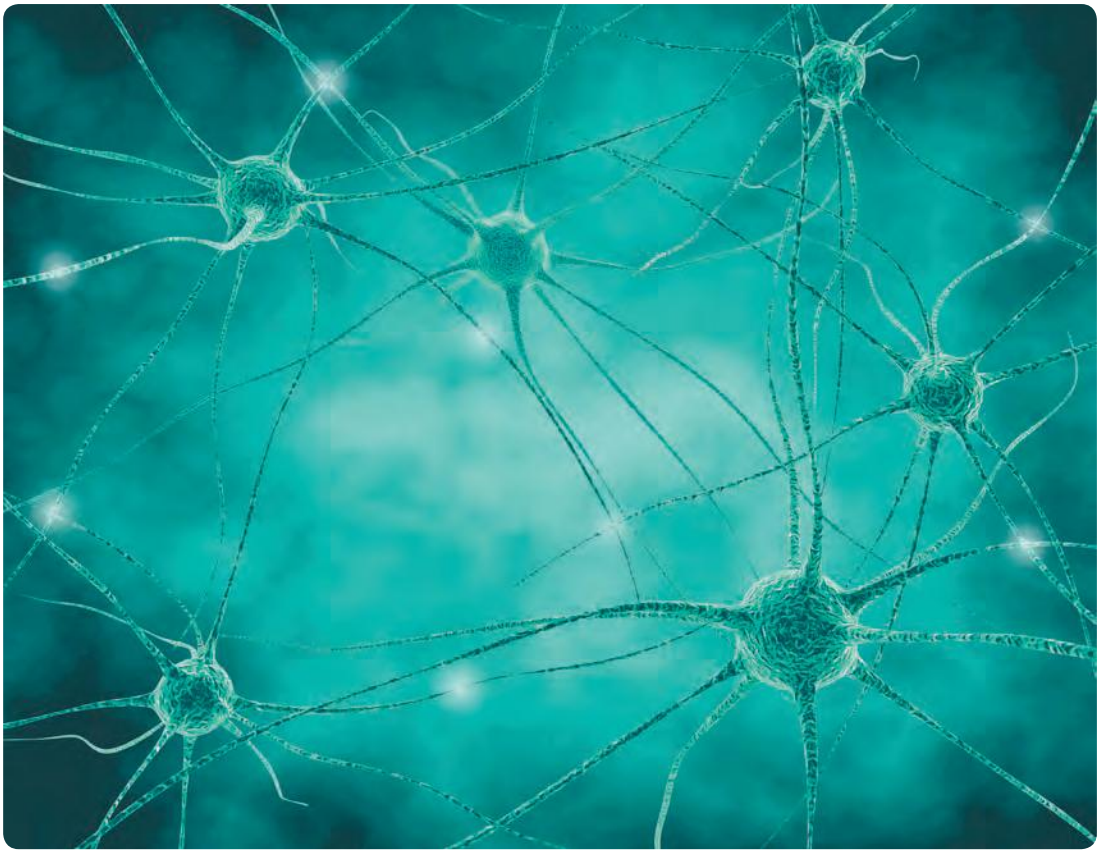
Shutterstock.com/Aldona Griskeviciene

FIGURE 2.28 The three different types of muscle tissue

Nervous tissue

Nervous tissue is made up of specialised nerve cells that are called **neurons**. Neurons have long projections from the body of the cell. When part of a neuron is stimulated, messages can be carried along these projections from one part of the body to another.

Nervous tissue is found in the brain, the spinal cord and the nerves.



Shutterstock.com/Illustration Forest

FIGURE 2.29 A three-dimensional image of nervous tissue

Activity 2.7
Looking at tissues

Key concept

Similar cells are arranged into tissues that carry out a common function. The different types of tissue are epithelial, connective, muscular and nervous.

Organs

Organs are body structures that are made up of two or more types of tissue. The tissues of an organ work together to carry out a particular task. For example, the heart is mostly muscle tissue, but it is covered and lined with epithelium. It also contains nervous tissue to make the muscle contract. All these tissues work together to pump the blood, which is a connective tissue.

Organs are distinct structures that usually have a recognisable shape. For example, you are probably familiar with the shape of the heart, the stomach, the lungs or the brain, all of which are organs.

Some large organs have smaller organs within them. The skin is the largest organ in the body and within it are many smaller organs, such as the sweat glands, nerves, hair and nails.

Systems

The various organs are organised into body systems, sometimes called organ systems. A **system** is a group of organs that work together to carry out a particular task. For example, the role of the digestive system is to break down food and to absorb it into the blood. Some of the organs that work together to allow the digestive system to carry out these tasks are the mouth, stomach, intestines and liver.

The main systems of the body and their functions are listed in Table 2.2.

TABLE 2.2 The major systems of the body and their functions

BODY SYSTEM	MAIN FUNCTION
Digestive	Ingestion, breakdown and absorption of food
Respiratory	Intake of oxygen and removal of carbon dioxide
Circulatory	Transport of nutrients, oxygen and wastes to and from cells
Excretory	Removal of wastes
Nervous	Detection of changes in the environment and coordination of body activities
Endocrine	Regulation and coordination of many body functions
Skeletal	Support and protection of body parts
Muscular	Movement and support
Immune	Protection against infection by micro-organisms
Reproductive	Production of new individuals

In addition to the major functions of systems listed in Table 2.2, most of the systems have secondary functions. For example, the bones of the skeletal system store minerals and produce blood cells, as well as providing support and protection. The circulatory system is involved in protection against disease in addition to being the body's transport system.

Each of the systems will be discussed in more detail in the chapters that follow.

The organism

All the body systems work together to meet the needs of a functioning **organism**. No system can work in isolation; they all depend on each other. Heart muscle needs oxygen, which the respiratory system supplies; brain cells need nutrients, which are absorbed by the digestive system and transported by the circulatory system, and so on.

Some organs are part of more than one system. The pancreas is part of both the digestive system and the endocrine system. Therefore, organs such as the pancreas further contribute to the integration of the parts of the body.

Activity 2.8
Touring the tissues

Questions 2.4

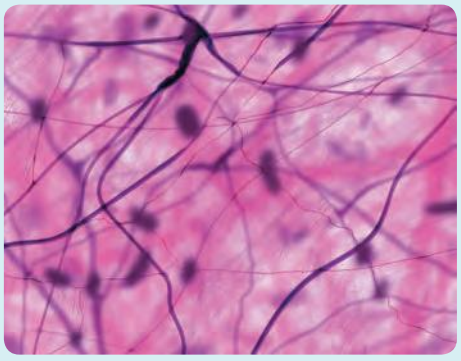
RECALL KNOWLEDGE

1. List the following in order from the lowest level of organisation to the highest: system, tissue, cell, organ.
2. What type of tissue is bone?
3. Define 'tissue'.
4. List three locations of epithelial tissue.
5. What are nerve cells called?
6. State the function of the circulatory system.

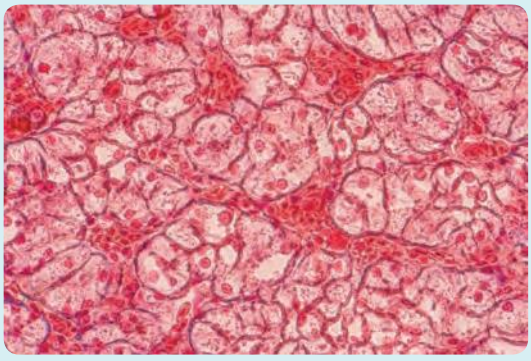
APPLY KNOWLEDGE

7 Classify each of the following tissues.

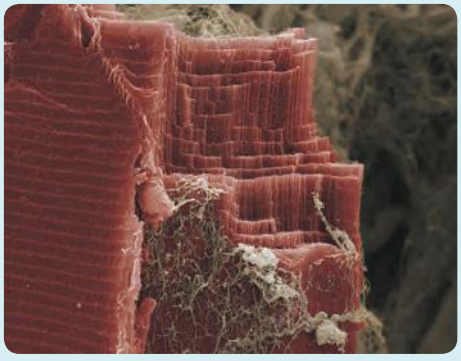
a



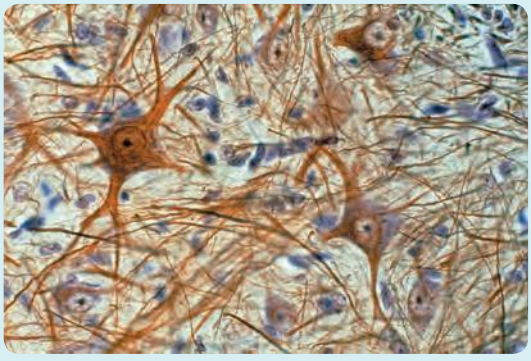
b



c



d



Clockwise from top left: Shutterstock.com/Angel Soler Gollonet; iStock.com/tonaquatic; Science Photo Library/Biophoto Associates; Science Photo Library/Anne Weston, Francis Crick Institute

8 Explain the difference between tissues and organs.

9 Explain why the cells in epithelial tissue are closely packed together.

9780170449090

CHAPTER 2 ACTIVITIES

ACTIVITY 2.1 Observing cells

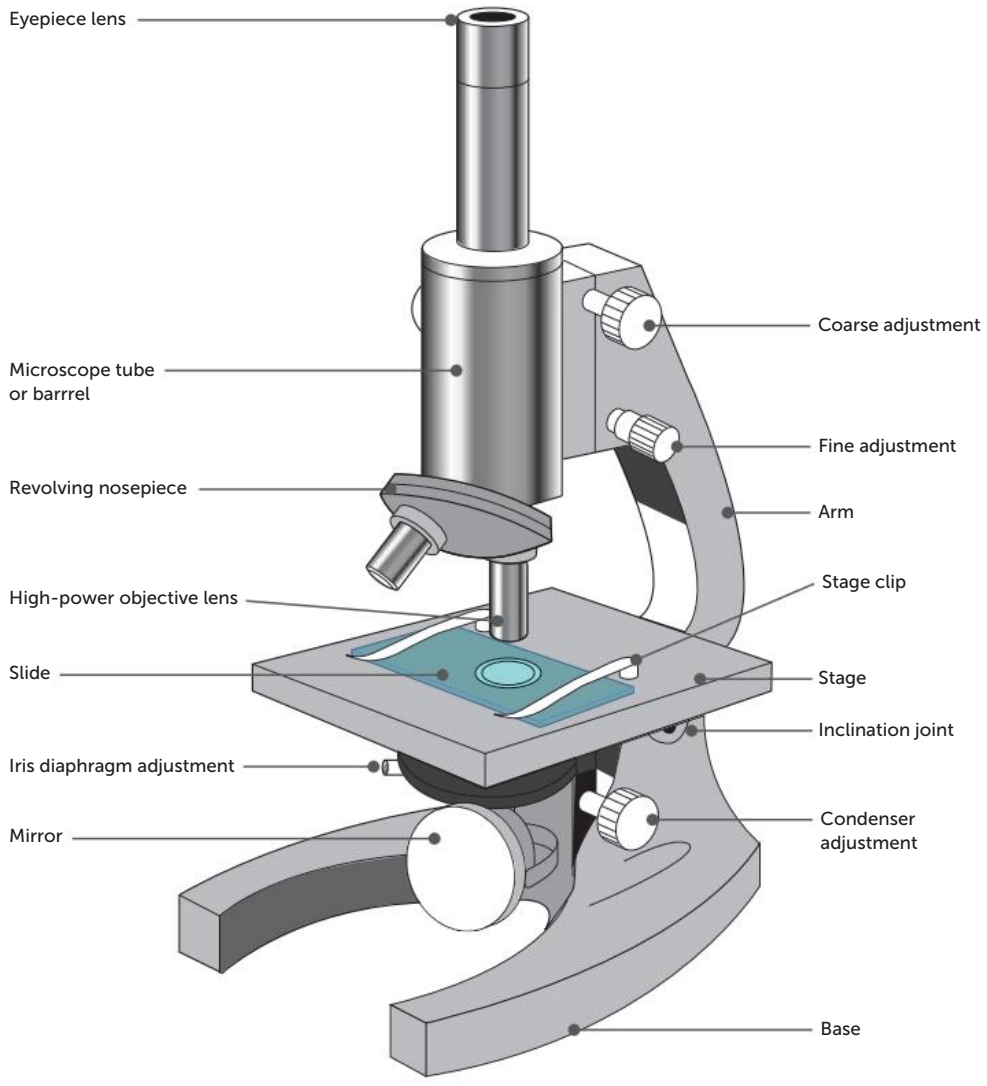
This activity will familiarise you with using a microscope to observe cells. You may have to work with a partner.

You will need

Microscope and microscope lamp; prepared microscope slides of cheek cells; minigrid or piece of millimetre graph paper

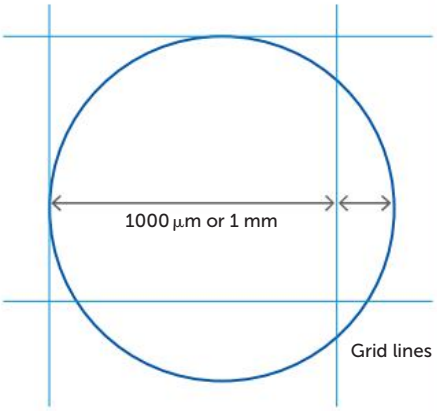
What to do

1 Use the illustration below to identify the parts of your microscope. Check:
   - **a** the number of objective lenses on your microscope and their magnification
   - **b** whether your microscope has a condenser with a condenser focus knob
   - **c** whether your microscope has a mirror or a built-in light source
   - **d** whether your microscope has an iris diaphragm or a wheel diaphragm.



How to use a microscope
Follow these instructions for using a microscope.

2 Rotate the nosepiece of the microscope so that the low-power objective lens (the shortest one) is in line with the body tube. It should click into position. If the microscope has a mirror, look through the eyepiece and move the mirror so that it reflects light up through the opening in the stage. If you are using a microscope lamp, use the concave side of the mirror.
3 While looking through the eyepiece, open and close the iris diaphragm or rotate the wheel diaphragm. If your microscope has a condenser, focus it up and down and observe any changes in light intensity.
4 Place a minigrid, or a slide with a piece of millimetre graph paper, on the stage. Lower the body tube until the objective lens almost touches the slide. While looking through the eyepiece, use the coarse adjustment to slowly *raise* the body tube until the specimen comes into view. With the fine adjustment, focus as sharply as possible. (*Never* focus down while you are looking through the eyepiece. The objective lens may hit the slide and break it, or the lens may be scratched.)
5 Move the slide so that one of the grid lines is on the very left of the field of view (see figure at right). As the grid lines are 1 mm apart, you can estimate the field of view using low power.
6 Remove the minigrid and place a prepared microscope slide of cheek cells on the stage of the microscope so that the cells you wish to examine are over the hole in the stage.
7 Focus the microscope on low power. Adjust the diaphragm so that you can see the maximum amount of detail. Note that you can often see more detail with a reduced light intensity, especially if the cells are almost transparent.
8 Turn the revolving nosepiece so that the high-power objective lens is in line with the barrel. If you do this carefully, the microscope should remain in focus or almost in focus.
9 Identify any structures that you can see in the cheek cells.



Studying your observations

1 What happens to the light intensity when you adjust the iris diaphragm or wheel diaphragm?
2 How does focusing the condenser affect the light intensity?
3 Which way does the image move when you move the slide on the stage to the right?
4 When you move the slide towards you, which way does the image move?
5 Compare what you can see with high power and low power. On which magnification do you see more of the specimen?
6 On which magnification is the image brighter?
7 Multiply the magnification of each of the objective lenses by the magnification of each of the eyepieces. List the magnifications that are possible with your microscope.
8 What was the field diameter on low power?
9 Estimate the diameter of the cheek cells in millimetres on the prepared slide.
10 One millimetre equals 1000 micrometres (μm). What is your estimate of the diameter of an average cheek cell in micrometres?
11 Draw a large, labelled diagram showing one or two cheek cells.
12 The cheek cells that you observed had been stained. What is the advantage of staining cells?

ACTIVITY 2.2 Making a model of a cell

Make a model of a cell showing all, or most, of the structures described in Figure 2.2 (on page 27). Use any materials you like to show the relative sizes and approximate shapes of the structures. Some ideas are lollies or other food items, household items, 3D printing, craft items or a virtual model. Label each of the structures, or number them and provide a key to the numbers.

ACTIVITY 2.3 Making a model membrane

Make a model of a cell membrane to show the membrane structure.

Think about the various components that you will need to show in your model. Everyday items could be used to represent each component, or you could construct the shapes in some way.

Label all the structures that make up the membrane.

ACTIVITY 2.4 Investigating diffusion through a differentially permeable membrane

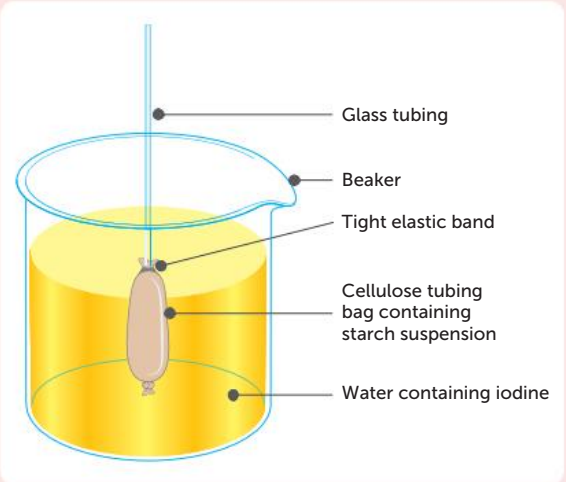
To get into and out of cells, substances must pass through the differentially permeable cell membrane. This activity will give you some understanding of the properties of differentially permeable membranes.

You will need

Cellulose tubing; glass tubing; 250 mL beaker; retort stand, clamp and boss; small elastic band; marking pen; starch suspension, 10%; iodine solution (iodine-potassium-iodide, I_2KI)

What to do

1. Cut a length of cellulose tubing about 12 cm long.
2. Tie a tight knot in the tubing near one end. Wet the tubing and open it so that it forms a bag.
3. Add starch suspension to the bag until it is nearly full.
4. Use an elastic band to attach the cellulose bag to the end of the glass tubing, as shown in the diagram at right. Ensure that your elastic band is very tight so there are no leaks.
5. Rinse the cellulose bag and glass tubing under the tap to remove any starch from the outside.
6. With a marking pen, mark the level of starch suspension in the bag.
7. Lower the bag into a beaker of water and hold the tubing erect using a retort stand and clamp.
8. Add iodine solution to the water outside the bag until the water is pale yellow.
9. Leave the set-up to stand for at least 40 minutes, and overnight if necessary.



Studying your results

Record any change in:

- the level of solution inside the cellulose bag or glass tubing
- the colour of the solution in the bag and the solution outside the bag.

Use your results to discuss and record answers to the following questions.

1. Do you have any evidence that any molecules passed from the beaker into the bag? Describe any such evidence.
2. Do you have any evidence that any molecules moved from inside the bag to the outside? Explain your answer.
3. Which has larger molecules: starch or iodine-potassium-iodide? Explain your answer. (You can estimate relative molecule size from the results of the experiment.)
4. Use the description of osmosis in this chapter to explain the changes that occurred in the experimental set-up.
5. If the cellulose bag containing starch suspension were a model of a cell, which part of the cell would be represented by the cellulose bag itself?
6. Predict what would happen if an isolated animal cell were placed in distilled water.

ACTIVITY 2.5 What size is it?

This activity will give you some practice in calculating the size of cells.

Figure 1 shows some cells as seen with the high power of a microscope.

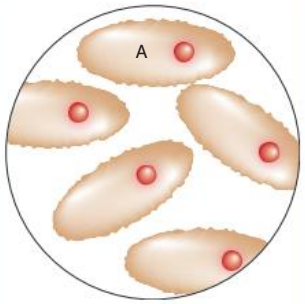


FIGURE 1 Cells seen with the high power of a microscope

1. If the field diameter is 0.5 mm, what is the approximate length and breadth of cell A in millimetres and in micrometres?
2. If the objective lens was changed from 40X to 10X, what would be the new field diameter?
3. How many cells the same size as cell A would fit end-to-end across the field with this new field diameter?
4. A student drew the cell shown in Figure 2. The actual length of the cell was 100 µm. What is the magnification of the student's drawing?
5. Estimate the length and width of the cell shown in Figure 3.

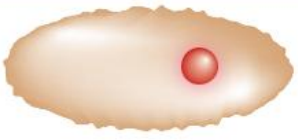


FIGURE 2 This cell is 100 µm long

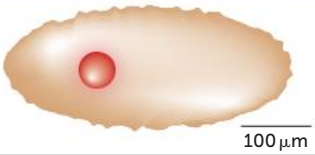


FIGURE 3 A cell

6. Estimate the diameter of the nucleus of the cell in Figure 2.
7. How many of the cells in Figure 3 would fit side-by-side across a field of view that has a diameter of 1.6 mm?

9780170449090

Developed by Southern Biological

ACTIVITY 2.6 Investigating surface area and volume

The relatively small size of cells allows molecules in and out of their membranes. If a cell becomes too large, the centre cannot be serviced efficiently. As the size of an object increases, the volume increases at a greater rate than the surface area. For a cell, this means that the efficiency of the exchange of materials across a membrane is reduced, and therefore the cell's ability to take in enough nutrients is also reduced. In addition, toxins may be retained for too long.

Using agar cubes with indicator, vinegar and some simple mathematics, we can see what effect a small increase in surface area has on volume.

Aim

To determine the relationship between surface area and volume ratio and its relationship to diffusion rates.
Time requirement: 45 minutes

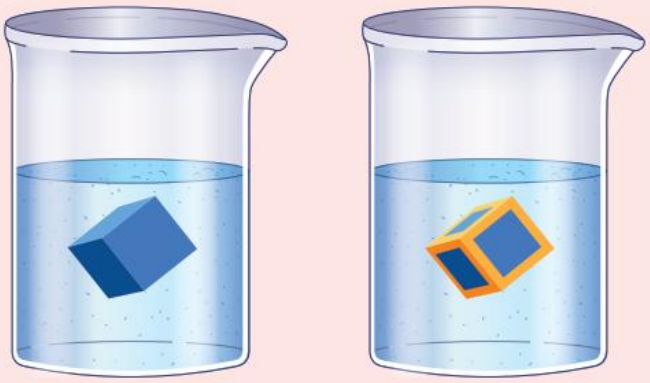
You will need

Prepared agar cubes impregnated with bromothymol blue indicator (1 × 1 cm^3, 1 × 2 cm^3, 1 × 3 cm^3); vinegar (acetic acid) 150 mL; 250 mL beaker; plastic or metal spoon; clock or timer; ruler; calculator; paper towel; disposable gloves

Risks

WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
Glass beaker may break or have chipped edges	Inspect and discard any chipped or cracked beakers, no matter how small the damage. Sweep up broken glass with brush and dustpan; do not use fingers.
Disposable gloves may pose allergy risk	Use a type of glove that removes allergy risk and is suitable for the chemicals being used.
Acetic acid may produce an irritant vapour	Ensure the investigation is performed in a well-ventilated space.

What to do



1. Propose a hypothesis for this investigation.
2. Put on disposable gloves and measure each cube in height (H), width (W) and length (L) to calculate surface area (SA) and initial volume (V_i).
3. Half-fill the beaker with vinegar, ensuring that the largest cube can be submerged, and place one cube of each size into the beaker.
4. After four minutes, remove the cubes, patting them dry with paper towel, and measure the portion of each that is still blue. Try to minimise the amount of time the cubes are out of the vinegar.
5. Replace the cubes and then repeat, leaving them in for four minutes at a time and measuring the dimensions of the blue portion after every four minutes for a total of 20 minutes.

6 Calculate the volume of the portion of the cube that is still blue (V_f) after each four-minute interval and the percentage of the whole cube that the vinegar has penetrated (%P).

Studying your results

Copy and complete the following tables.

Initial measurements

SA = surface area
V_i = initial volume

CUBE	HEIGHT (H) (cm)	WIDTH (W) (cm)	LENGTH (L) (cm)	SA (cm^2)	V_i (cm^3)	RATIO (SA/V) (cm^{-1})
A						
B						
C						

Measurements over time (complete one table for each cube)

P = penetration of the vinegar into the cube
V_i = initial volume (from table above)
V_f = volume of the part of the cube that is still blue

CUBE (X)	MEASUREMENTS OF THE CUBE THAT IS STILL BLUE					
TIME (min)	H (cm)	W (cm)	L (cm)	V_f (cm^3)	P ($V_i - V_f$) (cm)	% PENETRATION (P / V_i)
0						
4						
8						
12						
16						
20						

Discussion

1 Explain why the agar cubes change colour when placed in the vinegar solution.
2 Describe the relationship between the surface area and the rate at which diffusion occurs.
3 Create a graph of time in minutes (x-axis) against the %P (y-axis) of each cube. Comparing them all on one graph will demonstrate the differences in the rate of diffusion of each.

Conclusion

Summarise your findings. Comment on your hypothesis, explaining the advantages and disadvantages of cell size and including real-life examples.

Taking it further

Create a graph of initial surface-area-to-volume ratio (x-axis) against the time in minutes for the whole cube to change colour (y-axis) to demonstrate that as the ratio increases, the time taken to completely penetrate will decrease in a non-linear fashion.

9780170449090

ACTIVITY 2.7 Looking at tissues

In this activity, you will use your microscope skills to observe some of the cells and tissues described in this chapter.

You will need

Microscope and microscope lamp; prepared slides of tissues

What to do

When looking at cells on the prepared slides, remember that they have been stained to show up the structure of the cell and its contents. Many slides contain more than one tissue. If you are uncertain which cells to look at, check with your teacher.

I Epithelial tissues

Look at some epithelial cells scraped from the inside of the cheek. If you have already done Activity 2.1, you may be able to skip the cheek cells.

1. Draw a few of the cells and write a description of them.
2. Estimate the size of an individual cell.
3. Explain how the structure of the cells is suited to their function of providing a smooth lining to the inside of the cheek.

II Connective tissues

Look at a slide of cartilage.

4. In what ways does cartilage tissue differ from the cheek cells that you observed?
5. How is the structure of cartilage suited to its function of providing structural material that is firm but flexible?

Fat storage tissue is called adipose tissue. Examine a slide of adipose tissue.

6. Draw a few cells from adipose tissue and write a description of them.
7. Estimate the diameter of one adipose cell.
8. How is the structure of adipose tissue related to its function of fat storage?

III Muscular tissue

Examine a slide of skeletal muscle fibres.

9. Draw part of a skeletal muscle fibre and write a description of it.
10. Why are muscle cells known as fibres?
11. How many nuclei are present in the fibre that you have drawn?
12. Why is skeletal muscle sometimes known as striped or striated muscle?

Studying your observations

1. List the cells that you have seen in order from smallest to largest.
2. Write a brief paragraph explaining the relationship between the structure and function of tissues.

ACTIVITY 2.8 Touring the tissues

Imagine that you are the size of a red blood cell and you are taking a group of other tiny people on a tour of the tissues of the body. Describe what you would tell the tourists about the appearance, structure and function of the tissues that they would see.

9780170449090

CHAPTER 2 SUMMARY

- Living things are made up of cells whose activities allow the organism to function.

  Cells are made up of:
  - a cell membrane
  - a cytoplasm made up of the jelly-like cytosol and the organelles suspended within it
  - the nucleus
  - a cytoskeleton
  - inclusions.
- Each organelle has a specific role within the cell.

  Organelles include:
  - the nucleus, which controls the functioning of the cell
  - ribosomes, which are the site of protein synthesis
  - endoplasmic reticulum, which form channels and are involved in storing and transporting molecules
  - the Golgi body, which modifies and packages proteins
  - vesicles, which are membrane-bound sacs
  - lysosomes, which contain digestive enzymes
  - mitochondria, which are the site of cellular respiration
  - cilia and flagella, which are important in moving particles or cells.
- Cells need to take in their requirements, including glucose and oxygen, and remove wastes produced, including carbon dioxide.
- The cell membrane provides a physical barrier, controls the movement of substances into and out of the cell, and provides sensitivity and support for the cell.
- The cell membrane is described by the fluid mosaic model, with a phospholipid bilayer arranged with the hydrophilic heads on the outside and the hydrophobic tails on the inside.
- Substances are able to move across the cell membrane by various means.
  - With simple diffusion, they move with the concentration gradient directly across the membrane. If the substance is water, the movement is called osmosis.
  - Facilitated transport uses membrane proteins. Channel proteins allow facilitated diffusion, while carrier proteins allow facilitated diffusion or active transport. Facilitated diffusion is a passive process as the movement is with the concentration gradient, whereas active transport is an active process as it is against the concentration gradient.
  - Vesicular transport involves membrane-bound sacs called vesicles. Endocytosis brings substances into the cell, whereas exocytosis removes substances.
- The size of cells is limited by the surface area required to supply the needs of the volume. As the cell gets larger, the surface-area-to-volume ratio decreases, and therefore larger organisms are made up of many cells rather than one large cell.
- The structure of the body is organised into systems, which are made of organs, which are made of tissues, which are made of cells.

  Tissue is classified as follows:
  - Epithelial: lines and covers organs.
  - Connective: provides support and connection – includes bone, cartilage, tendon, ligaments and fat storage.
  - Muscular: tissue that is able to contract either voluntarily (skeletal muscle) or involuntarily (cardiac or smooth muscle).
  - Nervous: makes up the brain and nerves, carries messages around the body.
- Each body system is made up of organs that allow it to fulfil a specific function within the organism.

9780170449090

CHAPTER 2 GLOSSARY

Active process A process that involves the expenditure of energy

Active transport The use of energy to move substances, usually ions, across a cell membrane against the concentration gradient

Bilayer Two layers that make up a single membrane

Cardiac muscle The muscle that forms the wall of the heart

Carrier-mediated transport Transport of ions or molecules across a cell membrane by special carrier proteins

Carrier protein A protein that carries substances from one side of the cell membrane to the other

Cell membrane A membrane that forms the external boundary of a cell; also called the plasma membrane

Cell theory The principle that all living organisms are made up of cells and the materials produced by cells

Cellular respiration The chemical reactions that make energy available for the cell; also called tissue respiration or internal respiration

Channel protein A protein that allows ions, water and small molecules to pass through the cell membrane

Chromosome One of the 46 rod-like structures that appear in the nucleus of a human cell at the commencement of cell division and carry the genetic information, composed of nucleic acids and proteins

Cilia Hair-like projections on the outside of a cell; they beat rhythmically to move the whole cell or to move material across the cell surface; singular: cilium

Concentration A measure of the number of particles in a given volume

Concentration gradient A difference in concentration of a solution, often between the inside and outside of a cell; also called diffusion gradient

Connective tissue Tissue providing support for body organs

Cytoplasm The contents of a cell, excluding the nucleus; also called protoplasm

Cytoskeleton An internal scaffolding of protein fibres within the cytoplasm of a cell

Cytosol The liquid part of the cytoplasm of a cell

Deoxyribonucleic acid A molecule in the nucleus of a cell that determines the types of protein that a cell can make

Differentially permeable membrane Membrane that permits the passage of certain substances (usually small molecules) but restricts the passage of others (large molecules); also called a semipermeable, partially permeable or selectively permeable membrane

Diffusion The movement of particles of a liquid or a gas so that they are distributed evenly over the available space; usually taken to mean the net movement of ions or molecules from a higher to a lower concentration until they are evenly distributed

Diffusion gradient *see* concentration gradient

DNA *see* deoxyribonucleic acid

Endocytosis The process by which a cell takes in materials by enfolding and enclosing them; includes phagocytosis and pinocytosis

Endoplasmic reticulum A network of membranes forming channels through the cytoplasm of a cell; it is used for storage, support, synthesis and transport within the cell

Epithelium Tissue that forms the outer part of the skin and that lines hollow organs and ducts; a covering tissue; also called epithelial tissue; plural: epithelia

Exocytosis The process whereby the contents of the vesicles of cells are pushed out through the cell membrane

Extracellular fluid Fluid found outside the cells; it includes tissue fluid and blood plasma; also called tissue fluid

Facilitated diffusion The process whereby proteins allow the movement of substances through the cell membrane along the concentration gradient

9780170449090

Facilitated transport Proteins in the cell membrane allow molecules to be transported across the membrane

Flagella A long projection from a cell; often has the function of moving the cell

Fluid mosaic model The currently accepted model of cell membrane structure

Golgi body A structure in the cytoplasm of a cell consisting of a stack of flattened channels; it packages materials for secretion from the cell; sometimes called Golgi apparatus

Heart muscle *see* cardiac muscle

Homeostasis The maintenance of a relatively constant internal environment despite fluctuations in the external environment

Hydrophilic Water-loving

Hydrophobic Water-hating

Inclusion Chemical substances inside a cell in the form of granules or droplets

Involuntary muscle Muscle that is not under our conscious control; found in walls of internal organs; also called non-striated muscle or smooth muscle

Lipid Large organic molecules made up of fatty acids and glycerol

Lysosome A small sphere formed from a Golgi body; contains digestive enzymes

Matrix Non-cellular material between the cells of a tissue

Microfilament Protein fibres that move materials around the cytoplasm or move the whole cell

Microtubule Fine tubes that help to maintain the shape of the cell and hold the organelles in place

Mitochondrion A structure in the cytoplasm of a cell in which the aerobic stage of respiration occurs; plural: mitochondria

Muscle fibres The long cylindrical cells that make up skeletal muscles

Nervous tissue Tissue made up of nerve cells (neurons)

Net diffusion *see* diffusion

Neuron A nerve cell

Non-striated muscle *see* involuntary muscle

Nuclear membrane A membrane that separates the nucleus of a cell from the cytoplasm

Nuclear pore Gaps in the nuclear membrane

Nucleolus A structure within a cell's nucleus; involved in protein synthesis

Nucleus A large organelle in a cell; contains DNA

Organ A structure made up of different types of tissue working together

Organelle Structures within the cytoplasm of a cell, each with specific functions

Organism A living thing, with different body systems all integrated

Osmosis The diffusion of water molecules through a differentially permeable membrane from an area of higher water concentration to an area of lower water concentration

Osmotic pressure The pressure due to differences in concentration on either side of a differentially permeable membrane

Passive process A process that occurs without any input of energy

Passive transport The transport of substances across the cell membrane without the input of energy

Phagocytosis The process by which a cell surrounds, and takes in, solid particles

Phospholipid A lipid molecule that contains a phosphate group

Pinocytosis The process by which cells enfold, and take in, drops of liquid

Plasma membrane *see* cell membrane

Protein channel A pathway through a protein in the cell membrane that allows the passage of substances across the membrane

Ribosome Site of protein synthesis; located on the surface of rough endoplasmic reticulum in eukaryotic cells

Rough endoplasmic reticulum A form of endoplasmic reticulum that is covered with ribosomes that give it a rough appearance; involved in the synthesis of proteins

9780170449090

Simple diffusion The process of substances moving along the concentration gradient in a solution or across a semipermeable membrane, without the use of membrane proteins

Skeletal muscle Muscle attached to bones, under voluntary control; also called voluntary or striated muscle

Smooth endoplasmic reticulum A form of endoplasmic reticulum that is involved in the synthesis of lipids; is not covered with ribosomes

Smooth muscle *see* involuntary muscle

Solvent A substance, often water, in which a solute is dissolved

Striated muscle Muscle made up of dark and light bands; includes skeletal and cardiac muscle

System A group of organs that work together for a common function; also called an organ system

Tissue A group of cells that are similar in structure and function

Tissue fluid Fluid found in the spaces between the cells; also called interstitial fluid or extracellular fluid

Vesicle A small membrane-bound cavity in the cytoplasm of a cell, smaller than a vacuole

Vesicular transport The transport of materials into or out of a cell in membrane-bound sacs; also called bulk transport

Voluntary muscle Muscle under voluntary control

9780170449090

CHAPTER 2 REVIEW QUESTIONS

Recall

1 Name the organelles commonly found in human cells.

2 Describe the organisation of a cell.

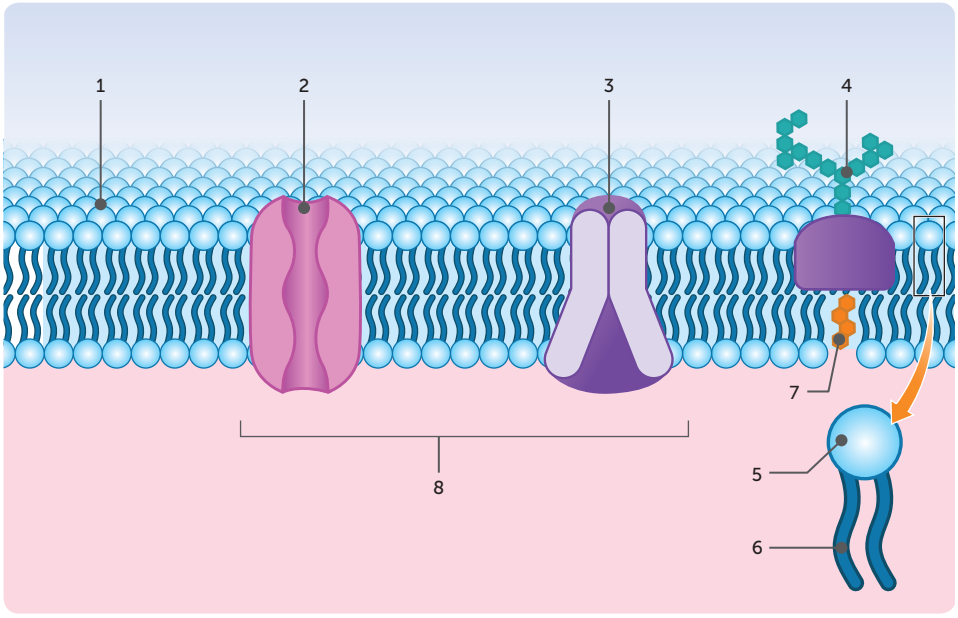
3 Describe the functions of the following organelles:
- a mitochondria
- b endoplasmic reticulum
- c ribosomes
- d nucleus.

4 What is a vesicle? Describe two ways in which vesicles can be formed.

5 Many cells have inclusions. Give two examples of inclusions.

6 List the substances that:
- a are required by all cells
- b have to be removed from all cells.

7 Label the diagram of the fluid mosaic model of the cell membrane.



8 Use a diagram to describe diffusion.

9 Define 'active transport' and provide an example that occurs in humans.

10 Describe the level of organisation within the human body.

11 Copy and complete the table below regarding the four different types of tissues.

TYPE OF TISSUE	FUNCTION OF THE TISSUE	LOCATION OF THE TISSUE IN HUMANS	DIAGRAM OF THE TISSUE

12 Choose two body systems and list the organs that are part of each of those systems.

9780170449090

Explain

13 Explain the difference between the cytosol and cytoplasm.

14 The nuclear membrane has large gaps in it. Explain the importance of these gaps.

15 Why are most cells microscopic?

16 Explain the relevance of concentration gradient.

17 Explain the role of proteins in transport across a cell membrane.

18 Explain the differences in function between the three types of muscle tissue.

19 Explain why, in the lungs, oxygen diffuses from the air into the blood but carbon dioxide diffuses from the blood into the air.

Apply

20 Unlike plant cells, animal cells have no cell wall. How is the shape of a human cell maintained?

21 Compare and contrast diffusion and osmosis.

22 Explain the importance of the structure of a mitochondrion.

23 Explain how the structure of the cell membrane makes it permeable to some molecules but not to others.

24 A red blood cell placed in distilled water swells up and bursts, but a red blood cell placed in sea water (about 3% salt) shrivels. Explain why this happens.

Extend

25 Predict how human cells would be different if the cell membrane was completely impermeable, rather than selectively permeable.

26 Explain how lysosomes and vesicles may work together.

27 Would you expect the cells of a large mammal, such as an elephant, to be larger than those of a small mammal, such as a mouse? Explain your answer.

28 Some experts do not regard the nucleus as an organelle. Suggest possible reasons why they believe that the nucleus should be classified separately from other organelles.

29 Patients who have suffered severe blood loss or dehydration have to be given large volumes of fluid. A fluid that is often given is a 0.9% solution of sodium chloride, known as normal saline. Why is saline solution given, rather than just plain water?

9780170449090

3 CELLS UNDERGO CHEMICAL REACTIONS

UNIT 1 CONTENT

SCIENCE INQUIRY SKILLS

- conduct investigations, including monitoring body functions; use microscopy techniques; and perform real or virtual dissection, safely, competently and methodically for the collection of valid and reliable data
- represent data in meaningful and useful ways; organise and analyse data to identify trends, patterns and relationships; qualitatively describe sources of measurement error, and uncertainty and limitations in data; and select, synthesise and use evidence to make and justify conclusions

SCIENCE UNDERSTANDING

Metabolism

- biochemical processes, including anabolic and catabolic reactions in the cell, are controlled in the presence of specific enzymes
- cellular respiration occurs, in different locations in the cytosol and mitochondria, to catabolise organic compounds, aerobically or anaerobically, to release energy in the form of adenosine triphosphate (ATP)
- for efficient metabolism, cells require oxygen and nutrients, including carbohydrates, proteins, lipids, vitamins and minerals
- enzyme function can be affected by factors including pH, temperature, presence of inhibitors, coenzymes and cofactors, and the concentration of reactants and products

Source: School Curriculum and Standards Authority, Government of Western Australia

9780170449090

3.1 METABOLISM

Although cells differ greatly in size, shape and the function they perform, they all carry out chemical processes that keep the organism alive.

All the chemical reactions that take place in cells, and therefore in the organism of which the cells are a part, are referred to as **metabolism**. Metabolism is made up of two different types of chemical reaction:

- Catabolic metabolism is the reactions in which large molecules are broken down to smaller ones. This process is known as **catabolism**. Digestion is an example of catabolism.
- Anabolic metabolism is the reactions in which small molecules are built up into larger ones. This process is also referred to as **anabolism**. Protein synthesis is an example of anabolism.

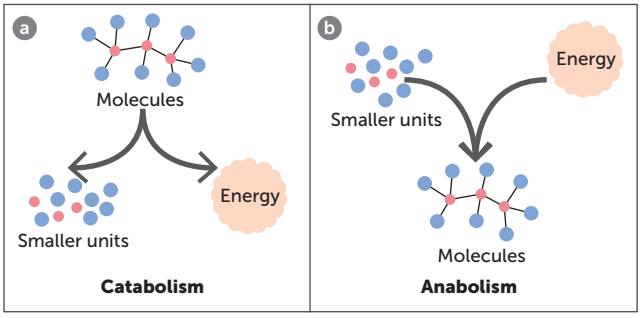


FIGURE 3.1
a Catabolism;
b Anabolism

Comparing catabolism and anabolism

Catabolic reactions release energy, whereas anabolic reactions require energy. Thus, metabolism is concerned with maintaining a balance between energy release and energy utilisation.

Nutrients

A **nutrient** is any substance in food that is used for growth, repair or maintaining the body; that is, any substance required for metabolism. There are six groups of nutrients: water, carbohydrates, lipids, proteins, minerals and vitamins.

Organic compounds

Organic compounds are molecules that have a carbon chain. They also contain a number of hydrogen atoms and may include atoms of oxygen, nitrogen and sulfur.

Carbohydrates are the main source of energy for cells. Simple sugars, particularly glucose, are used in cellular respiration to release energy. Complex carbohydrates, such as starch, are broken down to simple sugars.

CARBOHYDRATES

Carbohydrates always contain carbon, hydrogen and oxygen. There are always twice as many hydrogen atoms as oxygen atoms.

Monosaccharides are simple sugars or single-unit sugars; examples are glucose, fructose and galactose.

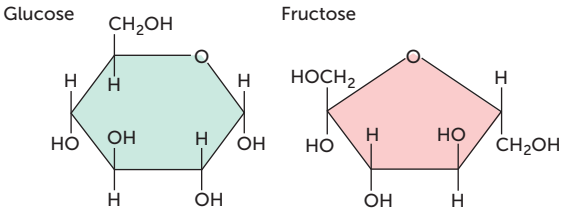


FIGURE 3.2 The structure of glucose and fructose

Disaccharides are two simple sugars joined together; examples are sucrose, maltose and lactose.

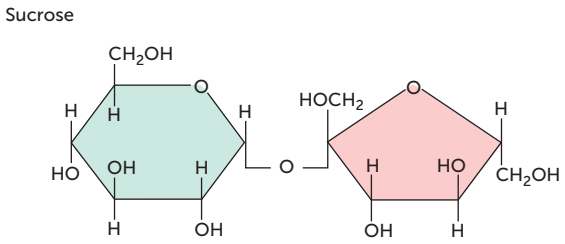


FIGURE 3.3 Sucrose is a disaccharide produced by a molecule of glucose and fructose bonding together

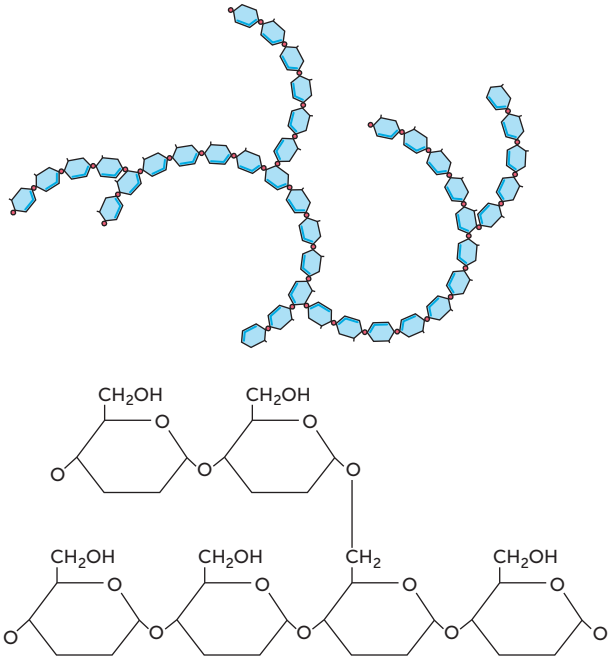
All carbohydrates contain atoms of carbon, hydrogen and oxygen; there are twice as many hydrogen atoms as oxygen atoms. Simple sugars are called **monosaccharides**. Glucose, fructose and galactose are examples of monosaccharides.

Simple sugars are able to join together to form larger molecules. **Disaccharides**, such as sucrose, maltose and lactose, are formed when two simple sugars join together.

9780170449090

FIGURE 3.4 Starch is a polysaccharide

Polysaccharides are large numbers of simple sugars joined together; examples are glycogen, cellulose and starch.



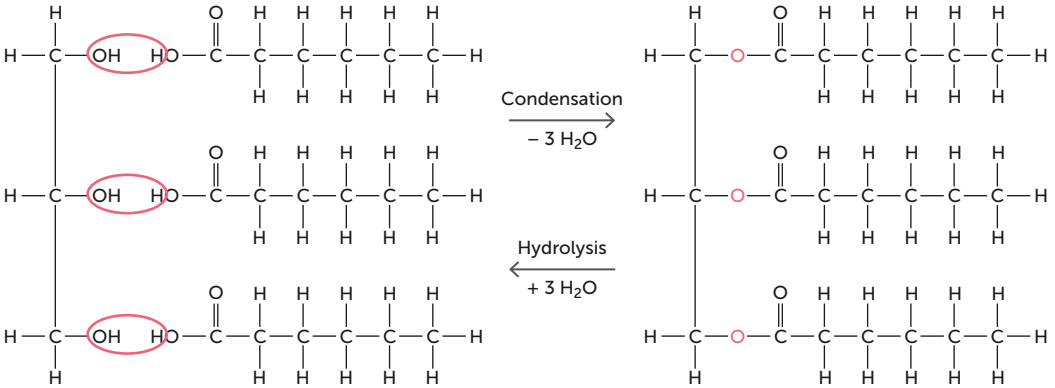
Carbohydrates provide energy for body cells.

Polysaccharides are larger carbohydrate molecules formed when many simple sugars join together. Glycogen, cellulose and starch are examples of polysaccharides.

Lipids include fats and oil and are another important energy source. They are broken down to fatty acids and glycerol. The glycerol can then enter the glycolysis pathway of cellular respiration and is broken down to release energy in a similar way to glucose. Other examples of lipids are phospholipids, which are important in the cell membrane, and steroids, including cholesterol and the sex hormones.

Each lipid molecule consists of one molecule of glycerol and one, two or three fatty acid molecules. The most common fat, including the fat that is stored in the body, is **triglyceride**, which is composed of glycerol and three fatty acid molecules.

FIGURE 3.5 Lipids are made up of glycerol and fatty acids



Proteins are organic compounds that are made up of many **amino acids**. With regard to metabolism, the most important proteins made are enzymes. Enzymes influence metabolism by controlling the chemical reactions that occur in the body. Proteins can also be used as a source of energy, but only if the supply of carbohydrates and lipids is inadequate.

An amino acid is a molecule that contains both an amino group and a carboxylic acid group. When two amino acids bond together, these two groups react to form a **peptide bond**, releasing a water molecule. There are 20 different amino acids found in proteins, each one differing in the structure of the side chain.

Proteins consist of 100 or more amino acids; their type and order are determined by the DNA that codes for the protein's production. Each protein has a characteristic shape due to the folding of the chain. Shorter lengths of amino acids include **dipeptides**, with two amino acids joined, and **polypeptides**, made up of more than 10 amino acids.

PROTEIN

Proteins always contain carbon, hydrogen, oxygen and nitrogen, and often sulfur and phosphorus. They are made up of large numbers of smaller molecules called *amino acids*. There are about 20 different amino acids; examples of amino acids are glycine, alanine, valine and glutamic acid. The bond that forms between amino acids is called a *peptide bond*; two amino acids joined by a peptide bond is a *dipeptide*.

Ten or more amino acids joined is a *polypeptide*. Proteins consist of 100 or more amino acids. Each protein's chain of amino acids is folded in a unique way. Proteins are important structural materials in the body. All enzymes are proteins, so proteins are involved in all the chemical reactions of the body.

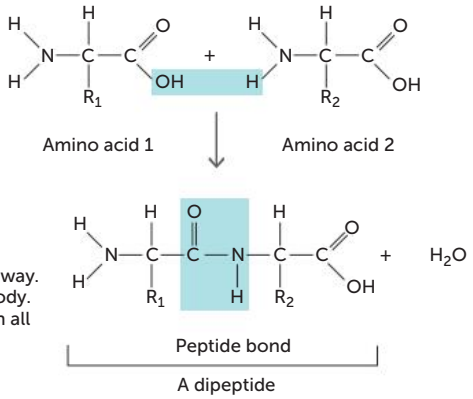


FIGURE 3.6 Two amino acids bond to form a dipeptide

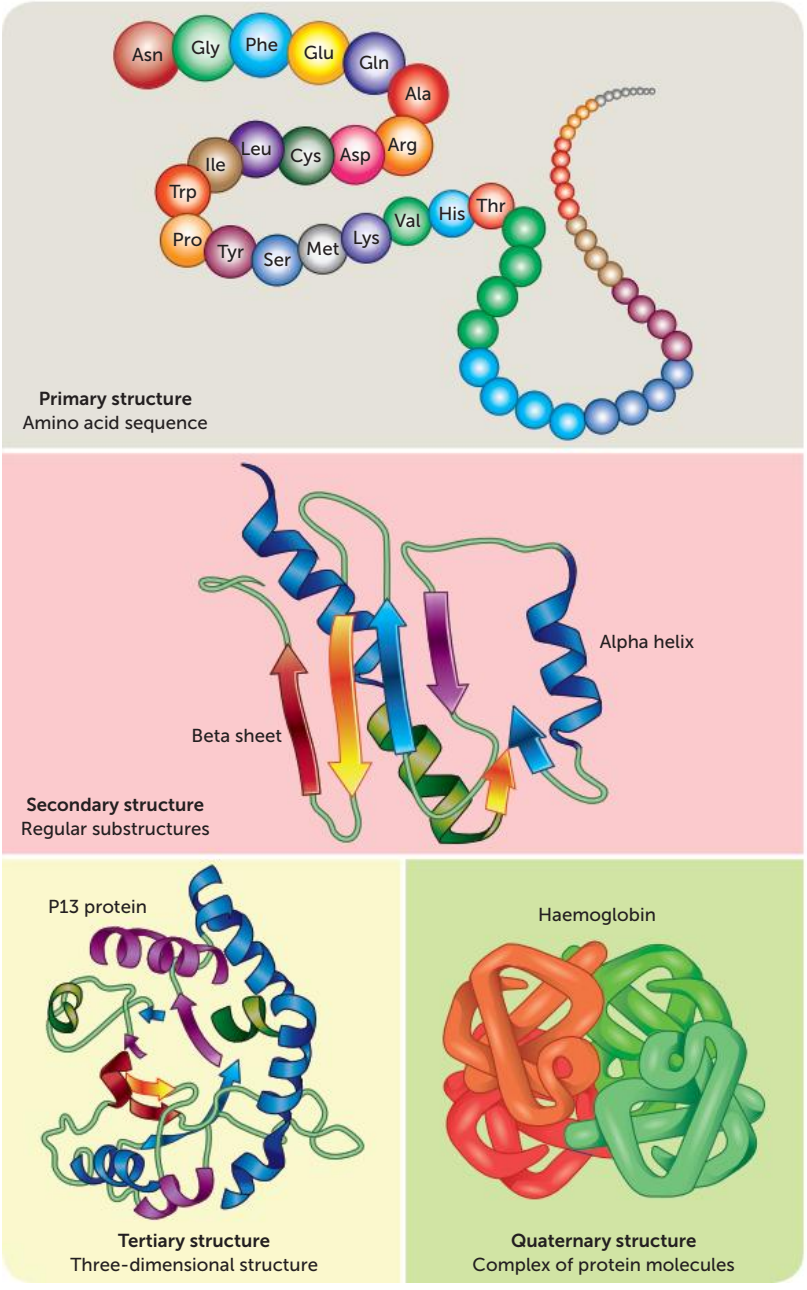


FIGURE 3.7 Proteins consist of a chain of amino acids that folds in a specific way to form a particular shape

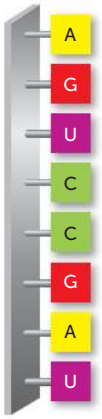
9780170449090

Other organic compounds include **nucleic acids** such as ribonucleic acid (RNA) and deoxyribonucleic acid (DNA). DNA consists of two chains of nucleotides that contain the sugar deoxyribose. It is the genetic material in the nucleus that stores inherited information. RNA is made up of a single strand of nucleotides that contain the sugar ribose. These molecules carry information from the DNA in the nucleus to the ribosomes for protein production.

FIGURE 3.8 a RNA is a single strand of nucleotides; **b** DNA contains a double strand of nucleotides

NUCLEIC ACIDS
Nucleic acids are very large molecules containing carbon, hydrogen, oxygen, nitrogen and phosphorus. They are made up of nucleotides, each of which contains a nitrogen base, a sugar and a phosphate. The two main kinds of nucleic acids are *ribonucleic acid, RNA, and deoxyribonucleic acid, DNA*.

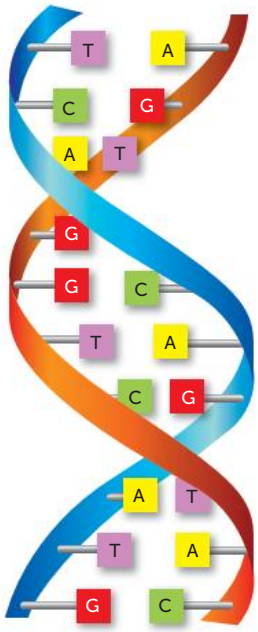
a **RNA** consists of a single chain of nucleotides that contain the sugar ribose.



RNA molecule

RNA carries information from the DNA in the nucleus to parts of the cell where proteins are made.

b **DNA** consists of two chains of nucleotides that contain the sugar deoxyribose.



DNA molecule

DNA is the genetic material in the nucleus that stores inherited information.

Inorganic compounds

Inorganic compounds are not based on a carbon chain. Most do not contain carbon atoms at all, but those that do, such as carbon dioxide, are small molecules. Some important inorganic compounds are water, minerals and vitamins.

- Water is important in metabolism because it is the fluid in which other substances are dissolved. Some of the cell's chemical reactions occur in water, and in others water molecules actually take part in the reaction.
- Minerals are important for metabolism because they may be a part of enzymes, may function as cofactors for enzymes, or may be a part of substances such as adenosine triphosphate (ATP) that are involved in metabolism.
- Vitamins act as coenzymes for many of the chemical reactions of metabolism.

WS

3.1 Nutrients and organic compounds

Questions 3.1

RECALL KNOWLEDGE

1. Define 'metabolism'.
2. Describe the structure of lipids.
3. Explain why amino acids bonding to form a protein is an example of anabolic metabolism.
4. Describe the role of water in chemical reactions.

APPLY KNOWLEDGE

5. Classify each of the following as either organic or inorganic: water, dipeptide, protein, minerals, polysaccharide, lipids, carbohydrates, nucleic acids, vitamins, monosaccharide, triglyceride.
6. Compare and contrast carbohydrates and proteins.
7. Conduct research to identify common foods that are high in simple sugars, complex carbohydrates, proteins and lipids.

9780170449090

3.2 ENZYMES AND METABOLISM

There are certain conditions that must be met for a chemical reaction to occur. The reacting particles need to collide with enough energy to break the bonds; this is the **activation energy**. The particles must also collide so that the correct atoms come into contact with one another.

At any given temperature, there is a certain proportion of particles that have enough energy to satisfy the activation energy. This proportion will increase when the temperature increases.

Chemicals called **catalysts** are able to decrease the amount of energy needed to break the bonds. This means that the activation energy will be lower, and more particles will have enough energy to react, making the reaction happen at a faster rate. Catalysts are particularly effective as they are not consumed during the reactions. Therefore, each catalyst particle is able to influence many reacting particles. In living things, catalysts are proteins called **enzymes**. Enzymes allow chemical reactions to occur at a fast-enough rate at body temperature for the body to function. Without enzymes, the reactions would be too slow.

Key concept

Enzymes are biological catalysts that are able to speed up chemical reactions by lowering the activation energy. They are not consumed or altered in the reaction.

The shape of the protein means that enzymes are specific for a particular reaction. The molecule on which an enzyme acts is called the **substrate**. Each enzyme will combine with only one particular substrate and is therefore involved in only one specific reaction. This occurs because the enzyme and its substrate have characteristics that are complementary to one another; that is, the enzyme and the substrate have a shape and a structure that allow them to fit together. The part of the enzyme molecule that combines with the substrate is called the **active site**. When the enzyme and substrate are combined, they are called an **enzyme–substrate complex**.

Two models are used to describe how enzymes function.

1 The *lock-and-key model* states that shape of the enzyme (the key) is always complementary to the shape of the substrate (the lock). Therefore, the two will fit exactly to form the enzyme–substrate complex.

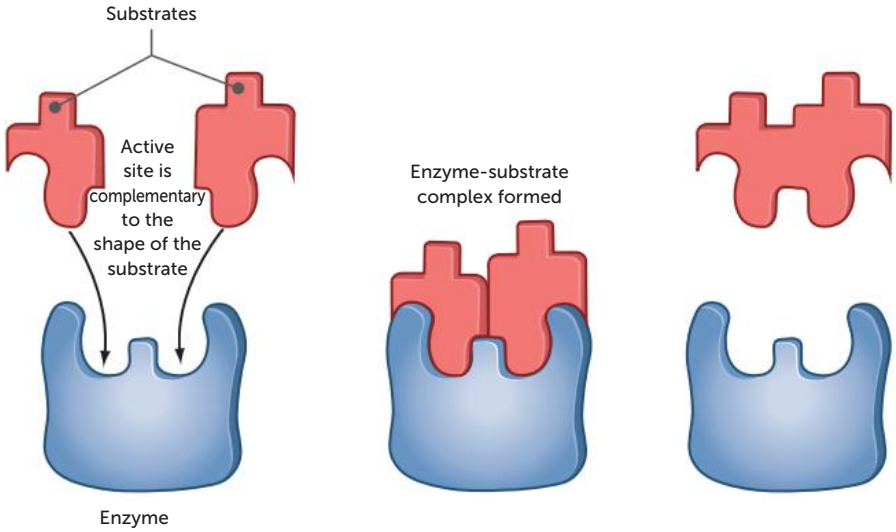


FIGURE 3.9 Enzyme action: Lock-and-key model

Lock-and-key model
This website provides diagrams modelling enzymes and substrates fitting like a lock and key.

9780170449090

2 The *induced-fit model* states that when the enzyme and substrate join, they form weak bonds that cause the shape of the enzyme to change, creating complementary shapes.

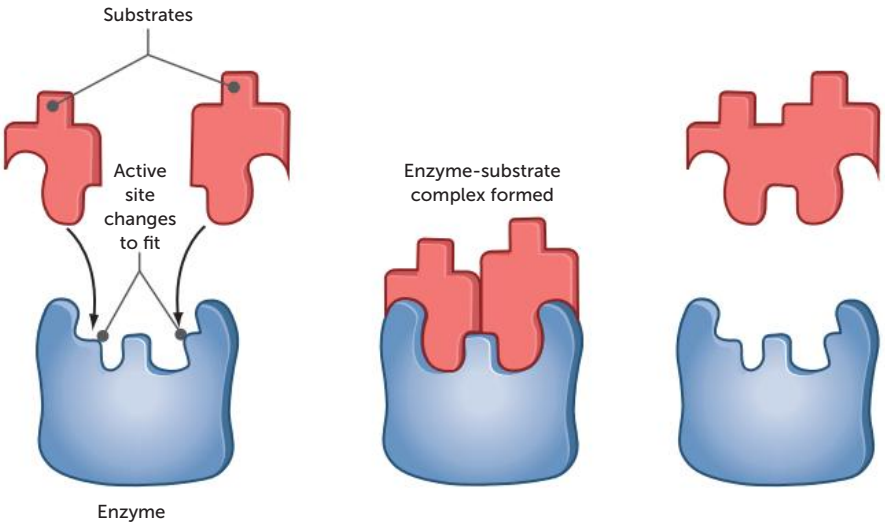


FIGURE 3.10 Enzyme action: Induced-fit model

Enzymes review activity
Enzymes
This website provides more detail on how enzymes work.

Factors affecting enzyme activity

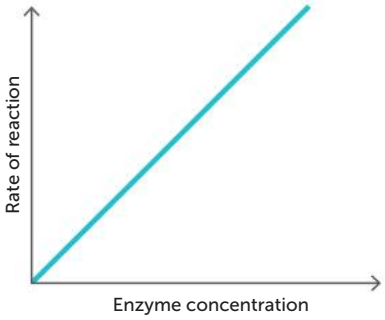


FIGURE 3.11 Graph showing the relationship between the concentration of the enzyme and the rate of the reaction

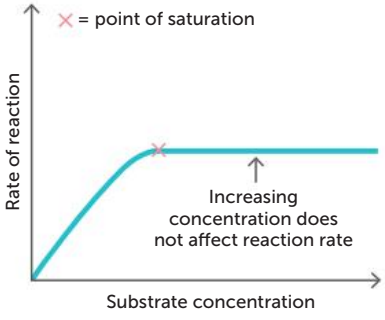


FIGURE 3.12 Graph showing the relationship between substrate concentration and the rate of reaction

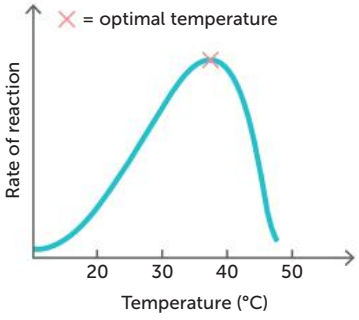


FIGURE 3.13 Enzymes have an optimal temperature for maximum impact on the rate of the reaction

A number of factors influence the activity of enzymes and the rates of chemical reactions in which they are involved.

- The higher the *concentration of enzyme*, the faster the rate of a chemical reaction because there are more enzyme molecules to influence reactants. By regulating the type and number of enzymes present, the body is able to control which reactions occur and the rate at which they proceed.
- Increasing *substrate concentration* also increases the rate of the reaction. This occurs because there will be more substrate molecules coming into contact with the enzyme molecules. However, increasing the substrate beyond a certain concentration will cease to have an effect because the active sites on all the enzyme molecules will be fully occupied.
- The *products of the reaction must be continually removed*, otherwise the rate of the reaction will slow because it becomes more difficult for the substrate molecules to make contact with the enzyme molecules.
- *Temperature* influences enzyme activity. The rate of most chemical reactions increases as temperature increases. This is true of most enzyme reactions but only within a limited temperature range. Because enzymes are proteins, beyond about 45–50°C their structure changes; they are **denatured**. As the shape of the enzyme is crucial for its functioning, denatured enzymes are inactive. The temperature at which an enzyme works best is called the *optimum temperature*. For most enzymes in the human body, this is 30°C to 40°C.
- Enzymes are very sensitive to the *pH* of the medium in which a reaction is taking place. Each enzyme has an optimum pH at which it will work most effectively.

9780170449090

- Many enzymes require the presence of certain ions or non-protein molecules before they will catalyse a reaction. Such substances are called ***cofactors***. Cofactors change the shape of the active site so that the enzyme can combine with the substrate. Without a cofactor the enzyme molecule is intact, but cannot function. Some cofactors are non-protein organic molecules. They are then called **coenzymes**. Many vitamins function as coenzymes.
- ***Enzyme inhibitors*** are substances that slow or even stop the enzyme's activity. Inhibitors may be used by cells to control reactions so that products are produced in specific amounts. Many drugs are enzyme inhibitors; for example, penicillin inhibits an enzyme in bacteria that is involved in construction of the cell wall.

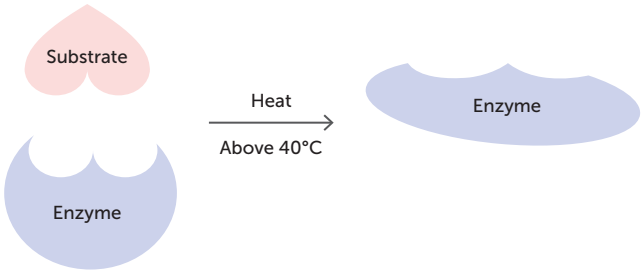


FIGURE 3.14 At high temperatures, the enzyme is denatured and the shape of the enzyme changes

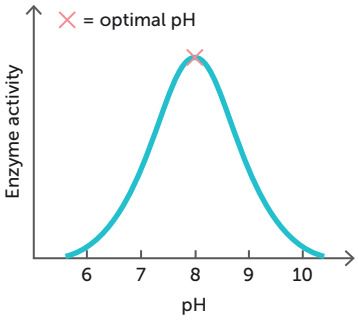


FIGURE 3.15 Enzymes have an optimal pH for maximum impact on the rate of the reaction

Key concept

The activity of enzymes is dependent on the shape and availability of the active site. Therefore, the effect of enzymes is influenced by temperature, pH, the concentration of both the substrate and enzyme, the removal of products, and the presence of cofactors, coenzymes and enzyme inhibitors.

Activity 3.1
Investigating the effect of temperature on trypsin activity

Questions 3.2

RECALL KNOWLEDGE

1 List the factors that affect the activity of enzymes.
2 Use a diagram to demonstrate the lock-and-key model of enzyme activity.
3 Explain why an increased temperature can:
   a increase the rate of the reaction
   b decrease the rate of the reaction.

APPLY KNOWLEDGE

4 Explain the difference between a cofactor and a coenzyme.
5 Lipase is an enzyme that catalyses the breakdown of lipid molecules. Would lipase be able to break a protein down into smaller peptides? Justify your answer.
6 Tay-Sachs disease is a genetic disorder where the enzyme hexosaminidase A is not produced. Without the enzyme, a fatty substance builds up on neurons, causing a degeneration of the central nervous system. Use this information to discuss the importance of enzymes.

3.3 CELLULAR RESPIRATION

Cellular respiration is one of the most important metabolic processes in any cell. It is the process by which organic molecules, taken in as food, are broken down in the cells to release energy for the cell's activities – activities such as the movement of the cell, uptake of materials from the surroundings, or production and secretion of new chemical compounds.

The term 'respiration' is often used loosely to mean breathing, and so the chemical process of respiration is referred to as cellular respiration. The process occurs in every cell in the body, to supply each cell with the energy it needs in the form of ATP and heat.

9780170449090

Cellular respiration can release energy from glucose, amino acids, fatty acids and glycerol. However, the main food material utilised is glucose, and the discussion here will therefore be confined to the respiration of glucose.

Respiration can be summarised as an equation:

$$\underset{\text{Glucose}}{C_6H_{12}O_6} + \underset{\text{Oxygen}}{6O_2} \longrightarrow \underset{\text{Carbon dioxide}}{6CO_2} + \underset{\text{Water}}{6H_20} + \underset{\text{(ATP and heat)}}{\text{Energy}}$$

This summary makes respiration look like a simple reaction. However, the breakdown of glucose to carbon dioxide and water actually involves more than 20 separate reactions, which occur in a series, one after the other. At each step, an intermediate compound is formed, and each step is catalysed by a different enzyme. Small amounts of energy are released as the reactions proceed. In this way, the release of energy is controlled rather than happening all at once.

Key concept

Cellular respiration is a multistep process that releases energy from glucose.

Energy from cellular respiration

In the complete breakdown of glucose to carbon dioxide and water, about 60% of the available energy is released as heat. Cells cannot utilise heat energy, but it is important in keeping the body temperature constant. Heat is continually lost to the environment, so a continual supply of heat is necessary in order to maintain body temperature.

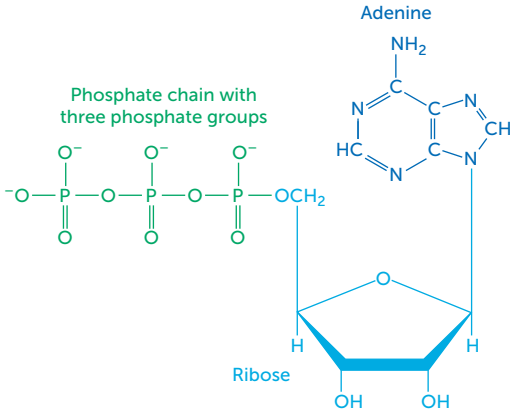


FIGURE 3.16
Structure of ATP

The remaining energy from cellular respiration is used to form a compound called **adenosine triphosphate (ATP)**. ATP is composed of:

- adenosine, which is made up of the nucleic acid base adenine and the sugar ribose
- three phosphate groups.

ATP is formed when an inorganic phosphate group is joined to a molecule of **adenosine diphosphate (ADP)**. The phosphate groups in ATP are joined by high-energy chemical bonds. Some of the energy from cellular respiration is stored in the bond between the ADP molecule and the third phosphate group. This bond is more easily broken than the bond between the first and second phosphate groups, allowing the energy to be released when needed. ATP can thus be used to transfer the energy released in cellular respiration to processes in the cell that require energy. The ADP formed when the energy is released can be reused to store some more of the energy from cellular respiration.

ATP
Learn more about ATP.

Key concept

Adenosine triphosphate (ATP) can be used to transfer energy between cellular respiration and processes in the cell requiring energy.

9780170449090

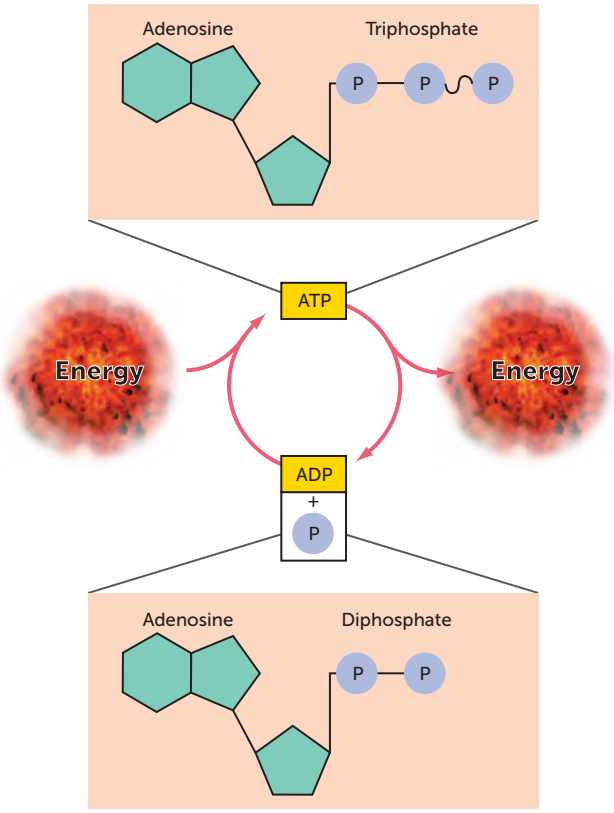


FIGURE 3.17 ATP stores energy; the energy is released when ATP breaks down to ADP

Glycolysis

The first phase in the breakdown of glucose does not require oxygen. It is called **glycolysis**, which means 'splitting glucose'. A glucose molecule is broken down, in a series of 10 steps, to two molecules of pyruvate. Sometimes this molecule is called pyruvic acid; however, the two substances differ slightly in their structure.

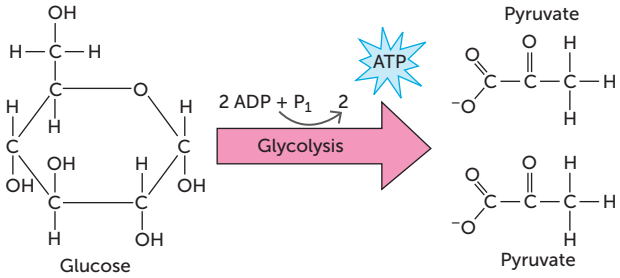


FIGURE 3.18 Summary of glycolysis

Pyruvate and pyruvic acid
Use this website to learn more about the difference between pyruvate and pyruvic acid.
Glycolysis

Anaerobic respiration

If no oxygen is available, the pyruvate produced in glycolysis is then converted to lactic acid by fermentation. The production of lactic acid from glucose is called **anaerobic respiration**, which means respiration without oxygen.

The fermentation stage of anaerobic respiration does not produce any additional ATP; however, the glycolysis of one molecule of glucose releases enough energy to convert two molecules of ADP to ATP. Therefore, anaerobic respiration allows cells to produce some energy in the absence of oxygen.

The enzymes required for anaerobic respiration are available in the **cytosol** of the cell; therefore, glycolysis and the conversion of pyruvate to lactic acid occur in the cytosol.

Anaerobic respiration is very important during vigorous physical activity, when the respiratory and circulatory systems are unable to supply muscle cells with enough oxygen to meet all the energy demands of the contracting muscles. In such circumstances, anaerobic respiration supplies the extra energy. This results in the accumulation of lactic acid in the muscles, and lactic acid may cause muscle pain.

9780170449090

Lactic acid from anaerobic respiration is taken by the blood to the liver, where it can be recombined with oxygen to form glucose and eventually glycogen. As this process requires oxygen, physiologists say that, when cells are respiring anaerobically, the body is incurring an **oxygen debt.** After vigorous exercise, one continues to breathe heavily for some time because the oxygen debt must be 'repaid' by converting lactic acid to glucose. The extra oxygen required after exercise may also be called **recovery oxygen**.

Key concept

Anaerobic respiration uses glucose to produce lactic acid and two ATP molecules in the absence of oxygen.

Aerobic respiration

The complete breakdown of glucose to carbon dioxide and water requires oxygen. The pyruvate produced from glycolysis is completely broken down to carbon dioxide and water. This is known as **aerobic respiration** – respiration requiring oxygen.

Aerobic respiration occurs in the mitochondria of the cell. Mitochondria are organelles constructed with a double membrane – an outer membrane that forms the shape of the organelle, and an inner membrane, called cristae, that is folded inwards. The enzymes for the reactions of aerobic respiration are attached to the internal membrane, so folding produces a large surface area on which the reactions of aerobic respiration can take place.

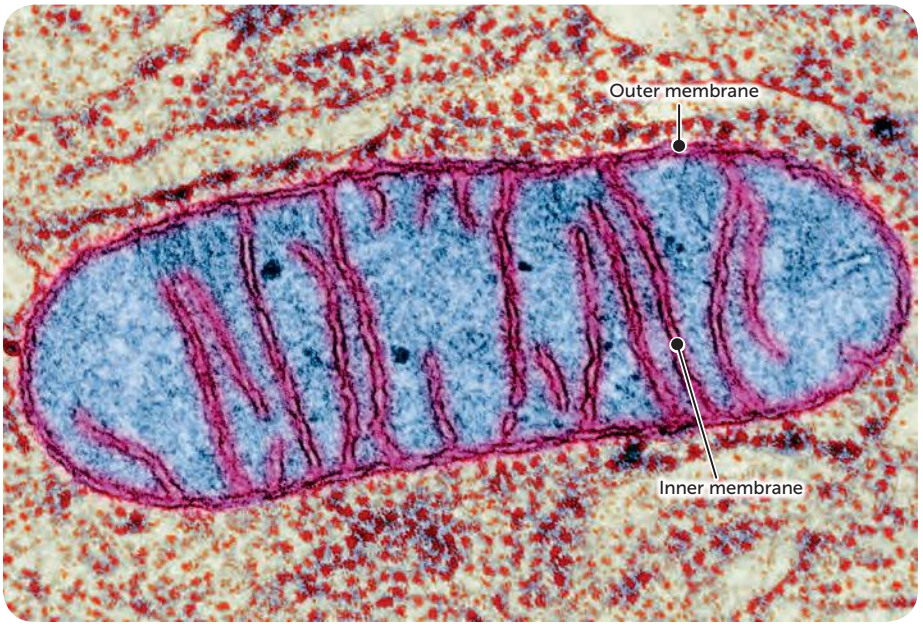


FIGURE 3.19 Transmission electron micrograph of a mitochondrion

To complete the breakdown of glucose, the two pyruvate molecules produced in glycolysis must enter a mitochondrion, where enzymes are available to allow the next series of reactions to occur.

1 For the pyruvate to enter the next pathway it is first converted to acetyl coenzyme A (acetyl CoA). To do this, a carbon dioxide molecule is removed from the pyruvate and the remaining two-carbon structure joins to coenzyme A. No ATP is produced during this process.

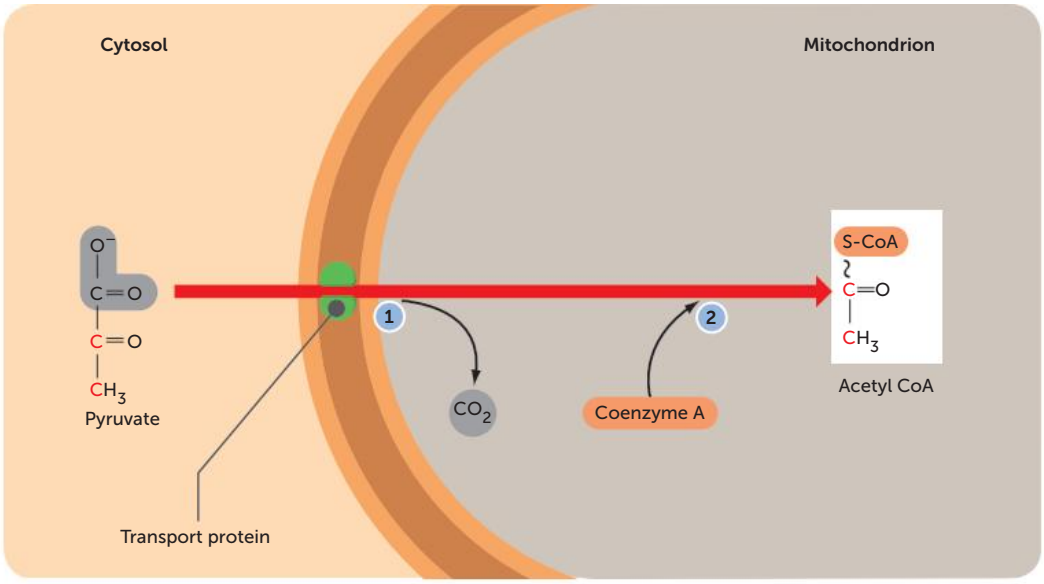


FIGURE 3.20
Pyruvate is converted to acetyl CoA

2 The acetyl CoA then enters the **citric acid cycle**, also known as the **Krebs cycle**. Here the carbon atoms in the acetyl CoA are released in carbon dioxide. For every acetyl CoA that enters the citric acid cycle, one molecule of ATP is also produced. This means that two ATP molecules are produced per glucose molecule.

3 The final stage of cellular respiration is the **electron transport system**; the only stage that uses oxygen. This stage is also called **oxidative phosphorylation**. Here electrons are passed between molecules, finally resulting in oxygen molecules forming water. There is some debate regarding the exact number of ATP molecules that are produced during this process. Estimates range between 26 and 34 molecules.

Thus, aerobic respiration of one molecule of glucose has the potential to generate up to 38 molecules of ATP – two from glycolysis, two from the citric acid cycle and up to 34 from the electron transport mechanism. This can be represented as:

$$C_6H_{12}O_6 + 6O_2 \longrightarrow 6CO_2 + 6H_2O$$
$$38\ ADP + 38\ P \qquad\qquad 38\ ATP$$

The processes of anaerobic and aerobic respiration are summarised in Figure 3.21.

Key concept

Aerobic respiration uses oxygen to convert glucose into carbon dioxide and water, producing up to 38 molecules of ATP per glucose molecule.

A yield of 38 ATP molecules from the energy contained in one molecule of glucose is the theoretical maximum. The actual ATP yield is often lower than this.

Because the reactions of aerobic respiration take place in the mitochondria, and because aerobic respiration releases about 95% of the energy needed to keep a cell alive, the mitochondria are often known as the powerhouses of the cell.

Cellular respiration

Cellular respiration information

Cellular respiration test
Test yourself on your knowledge of cellular respiration.

Activity 3.2
Investigating aerobic and anaerobic respiration during exercise

9780170449090

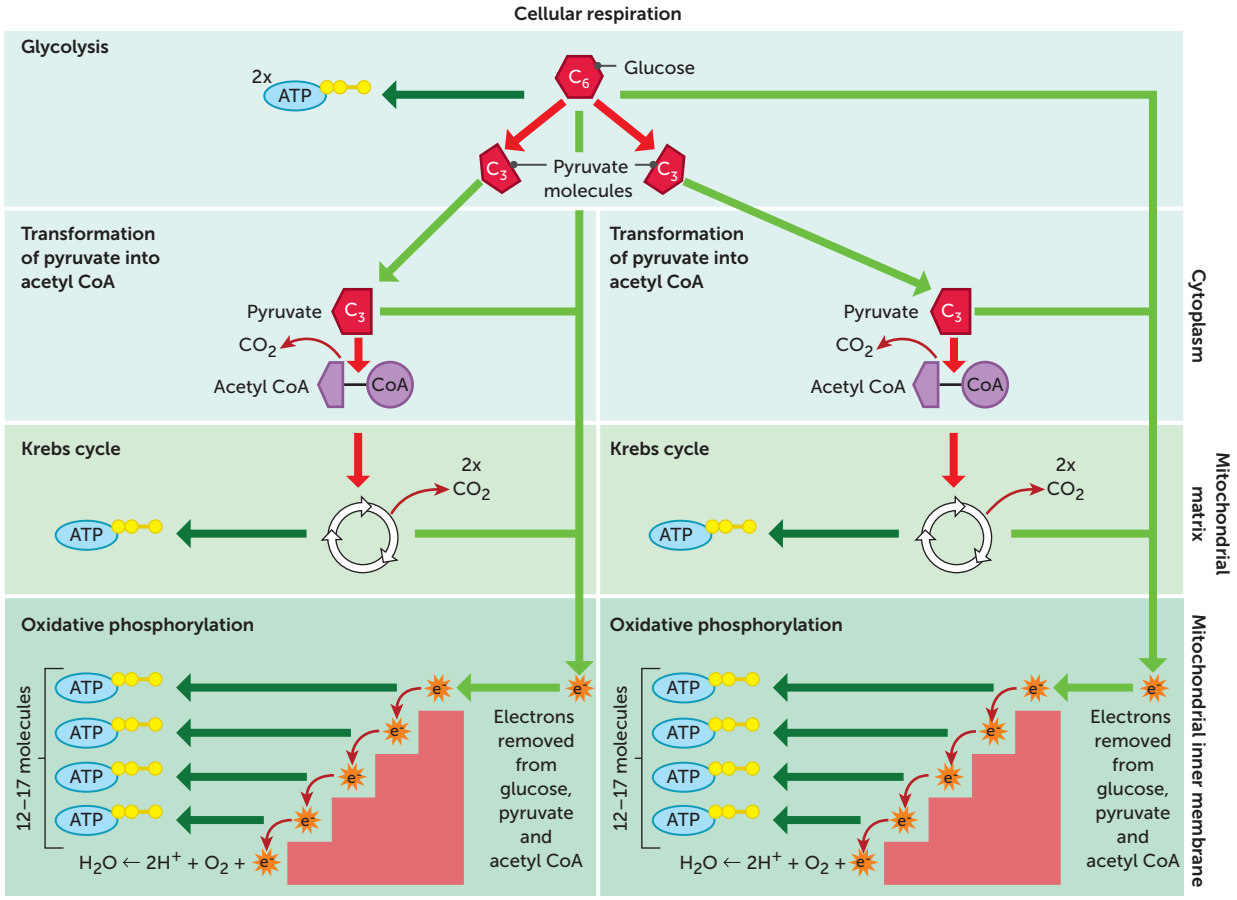


FIGURE 3.21 Summary of the processes of cellular respiration in a cell

Questions 3.3

RECALL KNOWLEDGE

1. Define 'cellular respiration'.
2. What does 'ATP' stand for?
3. Describe aerobic respiration.
4. Draw a flow chart to summarise the processes that occur during aerobic respiration.

APPLY KNOWLEDGE

5. Explain how ATP is able to store energy.
6. Describe the importance of the electron transfer system.
7. Construct a table to summarise the inputs and outputs for the stages of aerobic respiration, including glycolysis.

3.4 ENERGY USE BY THE CELL

Cells need the energy that is temporarily stored in the ATP molecule for a variety of processes. These are summarised in Figure 3.22.

Each of the chemical reactions involved in cellular processes produces a certain amount of heat. In cellular respiration, only about 40% of the energy released is incorporated into ATP; the other 60% is lost as heat. Therefore, energy must be continually consumed in the form of food to replace what is lost as heat and utilised for other purposes.

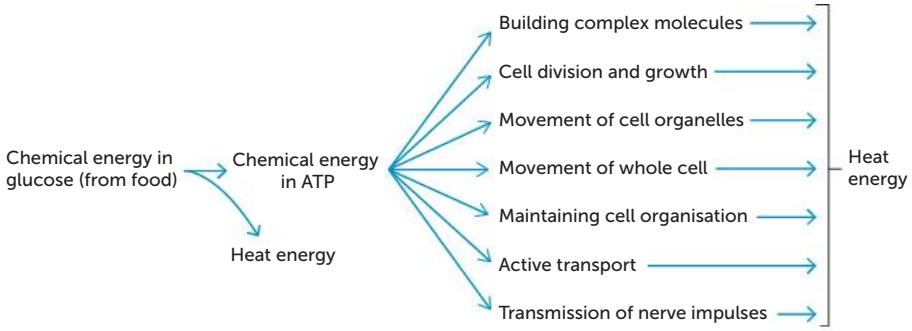


FIGURE 3.22 Uses of energy in the cell

The reactions of cellular respiration are catabolic; that is, they release energy as larger molecules are broken down into smaller ones. ATP may be used to transfer energy produced in catabolic reactions to anabolic reactions that require energy. For example, when lactic acid is recombined with oxygen in the liver to form glucose, or when glucose molecules are joined to form glycogen, the energy required comes from the breakdown of ATP to ADP. Similarly, energy for the build-up of proteins, lipids and other molecules is transferred from cellular respiration by ATP. Figure 3.23 models how ATP is able to transfer energy from one reaction to another.

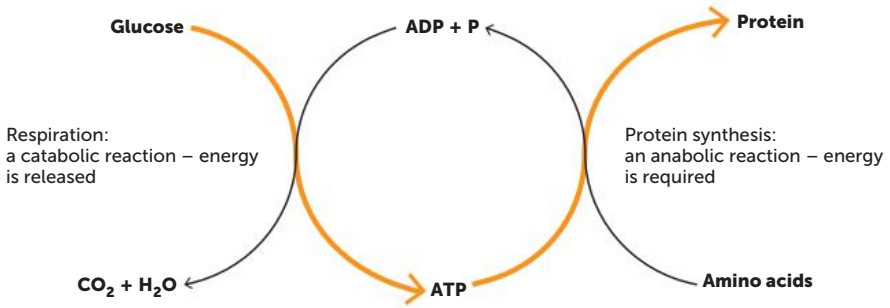


FIGURE 3.23 ATP transfers energy from reactions that release energy to reactions that require energy

Key concept

ATP is able to transfer energy from cellular respiration to other chemical reactions.

Questions 3.4

RECALL KNOWLEDGE

1. Define 'catabolic reaction' and 'anabolic reaction'.
2. List five ways that cells use energy.
3. Explain why energy is released when ATP forms ADP.

APPLY KNOWLEDGE

4. Approximately 60% of the energy produced during cellular respiration is released as heat. Discuss whether this energy is 'waste'.
5. Explain why cells that require a large amount of energy contain a lot of mitochondria.

9780170449090

CHAPTER 3 ACTIVITIES

Developed exclusively by Southern Biological

ACTIVITY 3.1 Investigating the effect of temperature on trypsin activity

Casein is a protein commonly found in mammalian milk. Casein is digested by trypsin, an enzyme which hydrolyses proteins into peptides, ready for other enzymes to cut them further down to their amino acids for use in the body. Trypsin works in the small intestine, after acid and pepsin in the stomach have commenced the work of breaking down the proteins. Casein is relatively hydrophobic, making it poorly soluble in water; however, when trypsin is added to a dilute solution of milk powder, the casein is digested and the solution goes clear.

Aim

To determine the optimal temperature for trypsin activity.
Time requirement: 45 minutes

You will need

1% trypsin solution; 3% solution of skim milk powder; pH 7 buffer solution; water bath; six test tubes; bungs or cork for test tubes; test-tube rack; clock or timer; marker; plastic pipettes; thermometer; disposable gloves

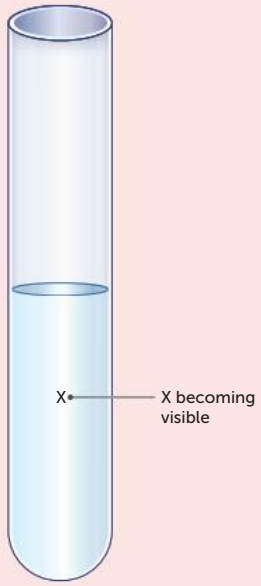
Risks

WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
Trypsin can cause allergic reactions in sensitive people	Be aware of any allergies.
Trypsin can be irritating to the skin and eyes on contact	Wear appropriate personal protective equipment at all times, including eye protection and gloves. Wash skin immediately if contact does occur.
Disposable gloves may pose allergy risk	Use a type of glove that removes allergy risk and is suitable for the chemicals being used.
Working with high temperatures	To prevent scalding, take care when working with water baths with water temperatures higher than 50°C. Do not touch the outside of the glass beaker.

What to do

1. Set the water bath to 20°C.
2. Collect and mark three test tubes with an 'X' on the glass halfway down the tube.
3. Using a pipette, add 10 mL of the milk powder solution to each of the three test tubes.
4. Collect another three test tubes, add 3 mL pH 7 buffer solution and then add 3 mL of trypsin solution.
5. Place all six test tubes in the 20°C water bath for 10 minutes. Ensure the six tubes are standing up.
6. Pour the trypsin and buffer solution from one test tube into the milk powder solution in another test tube.
7. To mix thoroughly, place a cork in the test tube and invert approximately five times.

8 Place the test tube in a test-tube rack and immediately begin a timer.
9 Record the time it takes for the milk solution to become clear in the table below as 'Test 1'. This may be achieved by measuring the time it takes for the 'X' to be visible through the solution.



10 Repeat this process (steps 6–9) for the remaining two test tubes and record the time for each of the three experiments in Table 1.
11 Calculate the average reaction time and record the result in Table 1.
12 Your teacher will now assign you one of four temperatures to test: 30°C, 40°C, 50°C or 60°C. As a class, you will test all four temperatures, pool your data and compare your results. Set the water bath to your assigned temperature. Once your water bath has reached the desired temperature, repeat the procedure (steps 2–10) to test how the reaction time is affected by different temperatures.
13 Record your data in Table 2, calculate the average time and share your results with the class.
14 Record the class average temperatures in Table 3. If one of the temperature variables was tested more than once, as in the 20°C control test, find the average among them.
15 Process your data and draw a graph using this data.

Studying your results

Copy and complete the following tables.

TABLE 1 20°C control test

SAMPLES	TIME FOR LIQUID TO BECOME CLEAR (min)
Test 1	
Test 2	
Test 3	
AVERAGE TIME:	

TABLE 2 Assigned temperature (Note your assigned temperature)

SAMPLES	TIME FOR LIQUID TO BECOME CLEAR (min)
Test 1	
Test 2	
Test 3	
AVERAGE TIME:	

TABLE 3 Class data

TEMPERATURE (°C)	AVERAGE TIME FOR LIQUID TO BECOME CLEAR (min)
20	
30	
40	
50	
60	

Create a graph of your results.

Discussion

1. What is your independent variable?
2. What is the range of your independent variable?
3. What is your dependent variable?
4. What are your control variables and how did you control them?
5. Why are all the test tubes left in the water baths for 10 minutes before the trypsin and milk are added together?
6. What are the advantages of taking an average of test samples as opposed to just one?
7. Why did the milk become clearer in this investigation?
8. You used a buffer solution in this investigation. What are buffer solutions used for?
9. What is the optimal temperature for trypsin activity?

Conclusion

Summarise your findings and discuss how the results are reflective of human body conditions.

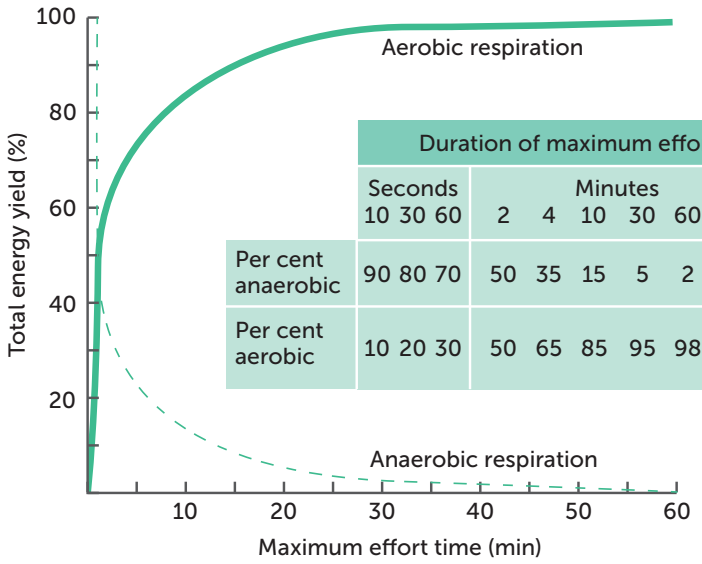
Taking it further

Change the variables of this investigation and test whether enzyme substrate concentration will cease to have an effect once the active sites of the enzyme are fully utilised.

ACTIVITY 3.2 Investigating aerobic and anaerobic respiration during exercise

During exercise, part of the energy required by the muscles comes from anaerobic respiration and part comes from aerobic respiration. The proportion of energy from each of the types of respiration depends on the nature of the exercise.

The graph and table below show the relationship between the duration of maximum effort and the proportion of energy derived from each of the two types of respiration. Use them to answer the following questions.



	Duration of maximum effort								
	Seconds			Minutes					
	10	30	60	2	4	10	30	60	120
Per cent anaerobic	90	80	70	50	35	15	5	2	1
Per cent aerobic	10	20	30	50	65	85	95	98	99

1. The world record for the 100 m sprint is less than 10 seconds. In a 100 m race, what proportion of a sprinter's energy would come from anaerobic respiration?
2. In a marathon race, what proportion of a runner's energy would come from anaerobic respiration?
3. At what duration of maximum effort would half an athlete's energy come from aerobic and half from anaerobic respiration? Can you suggest some sports in which maximum effort would come in bursts of that duration?
4. Name some sports or activities in which most of the energy would come from anaerobic respiration.
5. Name some sports or activities in which most of the energy would come from aerobic respiration.
6. Some observers noted that a sprinter who had just run 400 m in 50 seconds was breathing much more heavily than a runner who had just completed a marathon in 2.5 hours. Suggest why this would be so.

9780170449090

CHAPTER 3 SUMMARY

- Metabolism involves all the chemical reactions that take place in a cell. These may break large molecules down into smaller ones (catabolic metabolism) or use small molecules to make larger ones (anabolic metabolism).
- Nutrients from our food are used in metabolism.
- Carbohydrates, lipids, proteins and nucleic acids are all organic compounds, molecules based on a chain of carbon atoms.
- Minerals, water and vitamins are inorganic nutrients.
- Carbohydrates, the main source of energy for the cell, contain various numbers of simple sugars joined together.
- Lipids include fats, oils, phospholipids and steroids. They are composed of glycerol and fatty acids joined together. The most common lipid is triglyceride, which includes three fatty acids.
- Proteins are composed of amino acids joined by peptide bonds to form a chain. These chains are folded to form a particular shape.
- Nucleic acids are made up of chains of nucleotides. RNA has a single chain with the ribose sugar, whereas DNA has a double chain with the deoxyribose sugar.
- Enzymes increase the rate of chemical reactions by lowering the activation energy, which means that more particles have sufficient energy to react.
- Enzymes are specific for a particular substrate due to the shape of the active site. When they join, they form an enzyme–substrate complex.
- There are two models used to explain how enzymes work.
  - The lock-and-key model states that the active site and substrate have complementary shapes that allow them to fit together.
  - The induced-fit model states that when the substrate joins to the enzyme the enzyme changes shape so that the substrate fits.
- There are a number of factors that affect enzyme activity: the concentration of the enzyme, the concentration of the substrate, the removal of the product, temperature, pH, and the presence of cofactors, or enzyme inhibitors.
- Cellular respiration involves organic molecules being broken down to release energy.
- Glucose being broken down in cellular respiration is represented by:

  Glucose + Oxygen → Carbon dioxide + Water + Energy

  $C_6H_{12}O_6 + 6O2 \rightarrow 6CO_2 + 6H_2O + 38\ ATP$

- Adenosine triphosphate (ATP) is able to store energy in the bond between adenosine diphosphate (ADP) and the third phosphate group. This energy can then be released when needed.
- Glycolysis is the first stage of cellular respiration, where glucose forms two pyruvate molecules and two ATP molecules. This stage does not require oxygen.
- In the absence of oxygen, the pyruvate is converted to lactic acid. As no oxygen was needed, this is anaerobic respiration.
- In the presence of oxygen, the pyruvate undergoes aerobic respiration. This involves the pyruvate forming acetyl CoA before entering the citric acid cycle and then the electron transfer system. Up to 36 ATP molecules are produced during this process.
- Only 40% of the energy produced during cellular respiration is stored in ATP. The remaining 60% is released as heat.
- ATP produced during cellular respiration can be used in other cellular processes, including movement, active transport, nerve impulses, cell division and growth.

9780170449090

CHAPTER 3 GLOSSARY

Activation energy The energy needed to break the bonds of the reacting particles in a chemical reaction; the energy needed to start a chemical reaction

Active site The part of an enzyme molecule that combines with the substrate

Adenosine diphosphoate (ADP) The substance formed when the end phosphate group is removed from a molecule of adenosine triphosphate (ATP)

Adenosine triphosphate (ATP) A molecule that stores energy in cells; the energy is stored in the bond between the end phosphate group and the rest of the molecule

Aerobic respiration Respiration requiring oxygen

Amino acids Small molecules that join together to make proteins

Anabolism The process of combining small molecules to make larger ones; it requires energy; also known as synthesis

Anaerobic respiration Respiration that does not require oxygen

Carbohydrate Organic molecules that are the main source of energy for cells

Catabolism Chemical reactions that break down large organic molecules into smaller ones, with the release of energy

Catalyst A substance that lowers the activation energy of a reaction, increasing the rate of the reaction without being consumed

Cellular respiration The chemical reactions that make energy available for the cell

Citric acid cycle The series of reactions that occur in the mitochondria during aerobic respiration where acetyl CoA enters and carbon dioxide is released; also called the Krebs cycle

Coenzyme Non-protein organic molecules that are essential for the functioning of an enzyme

Cofactor The ions or inorganic molecules required by enzymes to catalyse a reaction

Cytosol The liquid part of the cytoplasm of a cell

Denature To change the molecular structure of a protein by heating, a change in pH, adding detergents or shaking

Dipeptide Two amino acids bonded together by a peptide bond

Disaccharide Two simple sugar molecules bonded together

Electron transport system A series of chemical reactions occurring in the mitochondria of a cell whereby energy from carrier molecules is transferred to ATP for storage

Enzyme An organic substance (usually a protein) that increases the speed of chemical changes without being altered or destroyed in the change; an organic catalyst

Enzyme inhibitor A substance that slows or stops an enzyme's activity

Enzyme–substrate complex The structure formed when an enzyme and a substrate combine

Glycolysis The breakdown of a glucose molecule to pyruvic acid; it releases energy to form two molecules of adenosine triphosphate (ATP)

Inorganic compound Not containing carbon (e.g. water, oxygen) or having small molecules (e.g. carbon dioxide)

Krebs cycle *see* citric acid cycle

Lipid Large organic molecules made up of fatty acids and glycerol

Metabolism All the chemical reactions occurring in a living organism

Monosaccharide A simple sugar molecule such as glucose or fructose

Nucleic acid Molecules containing many nucleotides forming a chain; includes ribonucleic acid (RNA) and deoxyribonucleic acid (DNA)

9780170449090

Nutrient Any substance in food that provides energy, is essential for growth, or assists in the functioning of the body

Organic compounds Substances that have large molecules and contain carbon, such as carbohydrates, amino acids, proteins, lipids and nucleic acids

Oxidative phosphorylation *see* electron transport system

Oxygen debt Extra oxygen required, in addition to the normal resting requirement, to remove the lactic acid produced during exercise

Peptide bond A bond formed between an amino group of one amino acid and the carboxyl group of another amino acid

Polypeptide Ten or more amino acids bonded together

Polysaccharide Many simple sugar molecules bonded together

Protein Very large organic molecule made up of amino acids

Recovery oxygen The extra oxygen needed to 'recover' after exercise

Substrate A molecule upon which an enzyme acts

Triglyceride The most common form of lipid, made up of glycerol and three fatty acids

9780170449090

CHAPTER 3 REVIEW QUESTIONS

Recall

1 Write the full names for ATP and ADP.

2 Describe the difference between breathing and cellular respiration.

3 What compounds are synthesised from:
- a glucose?
- b amino acids?
- c fatty acids and glycerol?

4 State the function of enzymes in the body.

Explain

5 Compare and contrast metabolism, catabolism and anabolism.

6 a Write a chemical equation that summarises cellular respiration.
- b Is the summary an accurate picture of what happens in cellular respiration? Justify your answer.

7 Explain the difference between aerobic and anaerobic respiration in terms of the:
- a quantity of energy released
- b reactions involved
- c location of the chemical reactions within the cell.

8 Explain what is meant by 'oxygen debt' or 'recovery oxygen'. How is an oxygen debt 'repaid'?

9 Explain why enzymes are substrate specific.

10 Explain why increasing the substrate concentration has no impact on the rate of the reaction beyond a certain point.

Apply

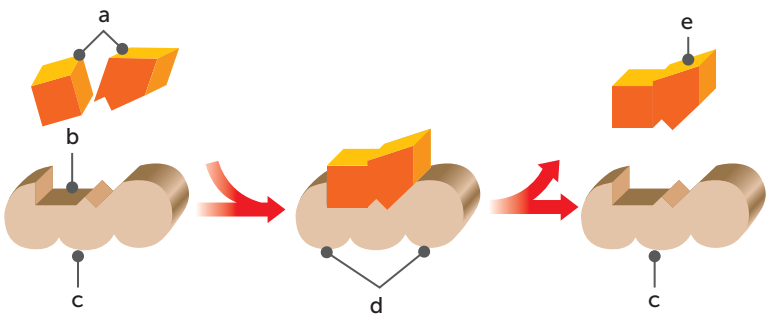
11 'Cellular respiration is vital for cellular functioning.' Explain this statement.

12 Use a diagram to summarise the relationship between ATP and ADP.

13 The law of conservation of energy states that energy can be neither created nor destroyed. If this is so, why do we need to continually take energy into the body in the form of food?

14 For each of the following processes, state whether the chemical reactions are anabolic or catabolic reactions:
- a protein synthesis
- b anaerobic respiration
- c formation of glycogen
- d aerobic respiration
- e formation of glucose from lactic acid.

15 The figure below is a model showing how an enzyme is involved in a chemical reaction. Which letter corresponds to the enzyme, substrate, active site, enzyme–substrate complex and product?



9780170449090

Extend

16 ACE inhibitors are medications that slow the activity of angiotensin-converting enzyme (ACE). Angiotensin is an enzyme that causes blood vessels to constrict. A person prescribed an ACE inhibitor would produce less angiotensin than usual. Suggest what medical condition/s ACE inhibitors could be used to control. Explain the reason for your suggestions.

17 Adolf Hitler and a number of high-ranking Nazi leaders committed suicide by taking cyanide. The cyanide ion travels to the mitochondria where it acts as an enzyme inhibitor for cytochrome C oxidase, an enzyme involved in the electron transport system. Explain why cyanide poisoning is fatal.

9780170449090

4 THE RESPIRATORY SYSTEM ALLOWS GAS EXCHANGE

UNIT 1 CONTENT

SCIENCE INQUIRY SKILLS

- identify, research and construct questions for investigation; propose hypotheses; and predict possible outcomes
- design investigations, including the procedure(s) to be followed, the materials required, and the type and amount of primary and/or secondary data to be collected; conduct risk assessments; and consider research ethics, including animal ethics
- conduct investigations, including monitoring body functions; use microscopy techniques; and perform real or virtual dissection, safely, competently and methodically for the collection of valid and reliable data
- represent data in meaningful and useful ways; organise and analyse data to identify trends, patterns and relationships; qualitatively describe sources of measurement error, and uncertainty and limitations in data; and select, synthesise and use evidence to make and justify conclusions

SCIENCE AS A HUMAN ENDEAVOUR

- lifestyle choices, including being active or sedentary, the use of drugs and type of diet, can compromise body functioning in the short term and may have long-term consequences

SCIENCE UNDERSTANDING

Respiratory system

- the exchange of gases between the internal and external environments of the body is facilitated by the structure and function of the respiratory system at the cell, tissue and organ levels
- the efficient exchange of gases in the lungs is maintained by the actions of breathing, blood flow and the structure of the alveoli

Source: School Curriculum and Standards Authority, Government of Western Australia

9780170449090

All cells in the body need oxygen for cellular respiration as well as a way to remove the carbon dioxide that they produce. In the lungs, oxygen is taken from air into the blood, and the blood transports the oxygen to the cells in all tissues of the body. In the tissues, the blood picks up carbon dioxide and takes it to the lungs where it is passed into the air. The circulatory and respiratory systems work together to ensure that all cells have a constant supply of oxygen and that carbon dioxide is continually removed from the cells. In this way, the amounts of oxygen and carbon dioxide in the tissues are kept relatively constant.

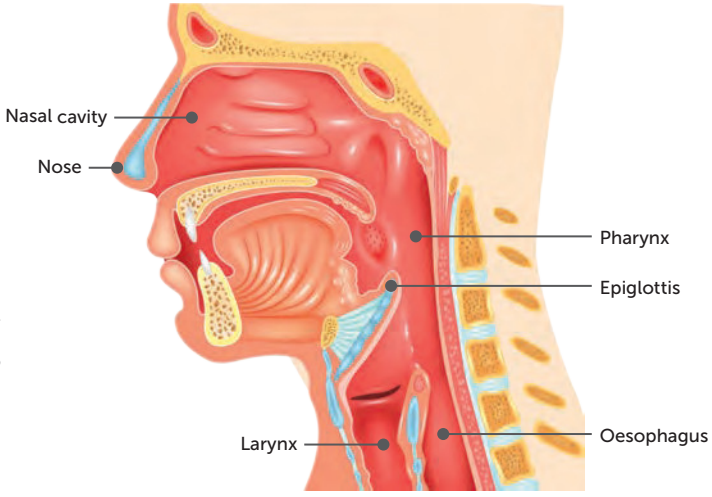
4.1 STRUCTURE OF THE RESPIRATORY SYSTEM

The organs of the **respiratory system** include the nose, through which air is taken in; the trachea, or 'windpipe', which branches into two tubes; the bronchi; and the two lungs.

Respiratory system
Explore the respiratory system at this website.

Nose, pharynx and larynx

Air enters the body through the mouth and nose. The lining of the nose and nasal cavity is convoluted and lined by mucous membranes. As the air passes over the membranes, it is warmed and humidified. There are also hairs and mucus lining the nose. These trap debris, preventing it from reaching the lungs.



Shutterstock.com/Teguh Mujiono

FIGURE 4.1 Anatomy of the nose, nasal cavity, pharynx and larynx

The **pharynx**, or throat, is the region from the nasal cavity to the top of the trachea and **oesophagus**. Air travels through it before being diverted into the trachea by the **epiglottis**, a flap of elastic cartilage. During inhalation the epiglottis covers the oesophagus, guiding the air into the trachea; when swallowing, the epiglottis covers the larynx, preventing food from entering it.

The **larynx** is a cartilage structure joining the pharynx and trachea. The larynx contains the **vocal cords**, which are mucous membranes that are able to vibrate as air passes over them. This is why the larynx is also known as the voice box.

Trachea

The **trachea** is also known as the windpipe because it carries the air into and out of the lungs. It is made up of C-shaped cartilage rings that hold the structure open. This ensures that air can always pass through it. At its base, the trachea splits into two branches, one branch taking air into each lung.

The epithelial lining of the trachea produces mucus, which is able to trap dust and debris. This prevents it from entering the lungs. Instead, the cilia that also line the trachea are able to move in a wave-like motion to take the mucus and debris up to the pharynx so that it can be swallowed and digested.

Bronchi

At the end of the trachea, the structure splits into two **primary bronchi,** one for each lung. These then split further into secondary bronchi which take the air into each lobe of the lung. The **secondary bronchi** continue to divide, forming **tertiary bronchi**.

The structure of the bronchi is very similar to the trachea, with C-shaped cartilage rings. As the bronchi get smaller, the cartilage is more spread out, with smooth muscle and elastin forming more of the structure. As in the trachea, cilia and mucus work together to trap and remove dust and other particles from the airways.

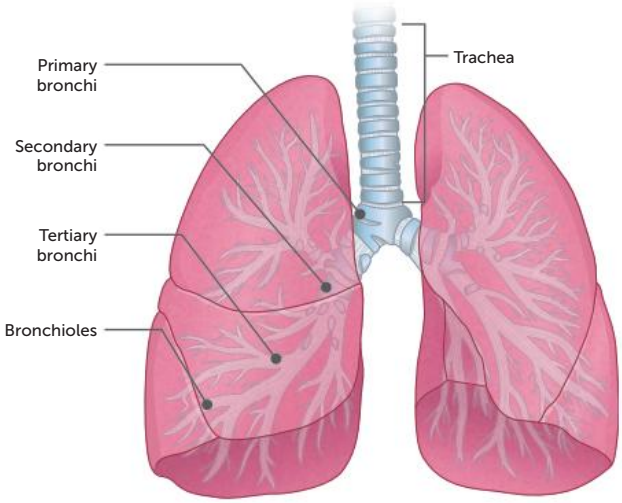


FIGURE 4.2 Trachea, bronchi and bronchioles

Bronchioles

When the tertiary bronchi divide, they form smaller airways called **bronchioles**, which continue to split until they end in millions of **terminal bronchioles**. Unlike bronchi, bronchioles do not contain cartilage; instead, they are made of smooth muscle and elastin. This allows the bronchioles to control the flow of air in the lungs, expanding when the body needs more oxygen. Cilia and mucus are also present in the bronchioles, protecting the lungs from contaminants.

Lungs

The two lungs take up the whole of the chest cavity, except for the space between them, called the mediastinum, that is occupied by the heart and blood vessels. Each lung is divided into lobes. The left lung has two lobes; the right lung has three. A membrane, called the **pleura**, covers the surface of the lungs (the visceral pleura) and also lines the inside of the chest (the parietal pleura). Between these two layers of membrane is a thin layer of **pleural fluid**, which holds the lungs against the inside of the chest wall and allows them to slide along the wall when breathing.

Inside the lungs the smallest bronchioles open into clusters of tiny air sacs called **alveoli**. Each alveolus has a wall that is only one cell thick and is surrounded by a network of blood capillaries. This is where gases move between the blood in the capillaries and the air in the alveoli. Therefore, the alveoli are the functional units of the lungs. This makes it possible for the alveoli to be the surface for gaseous exchange, allowing a net flow of oxygen to pass from the airways into the blood and carbon dioxide to pass from the blood into the airways.

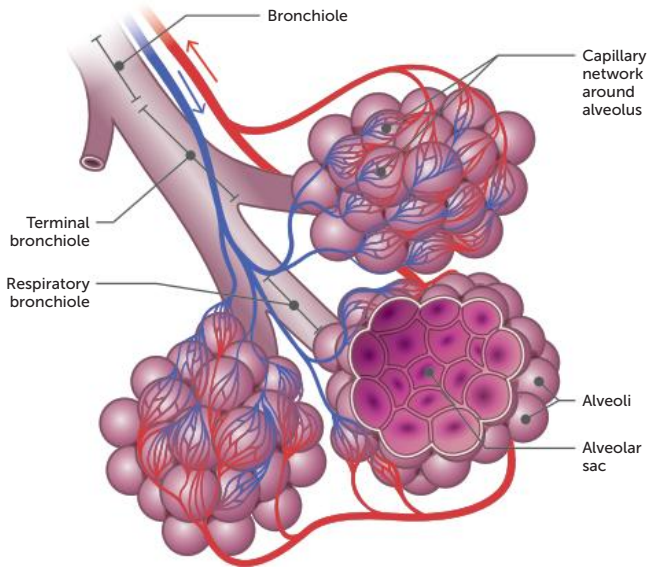


FIGURE 4.3 Structure of the alveoli

9780170449090

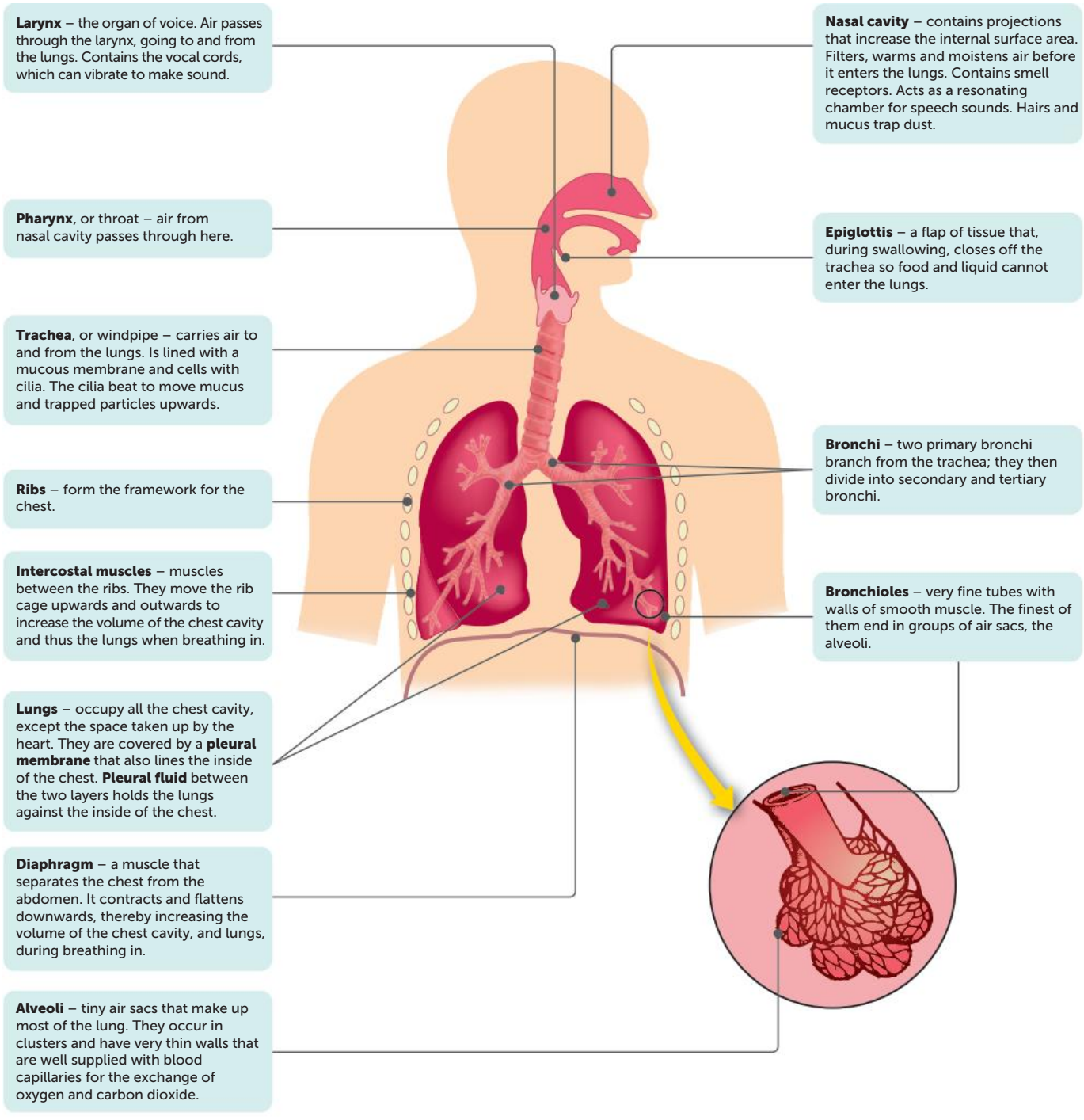


FIGURE 4.4 Overview of the respiratory system

Activity 4.1
Examining the structure of the lungs

WS

4.1 The respiratory system

Key concept

The structure of the respiratory system allows the efficient flow of air into and out of the lungs so that gas exchange can occur between the air and the blood.

Questions 4.1

RECALL KNOWLEDGE

1 List the structures that air will travel down, starting from outside of the body.
2 Describe the structure of the trachea.
3 Describe the function of the pleural fluid.
4 Describe the difference between a primary bronchus and a tertiary bronchus.

APPLY KNOWLEDGE

5 Explain the importance of the convolutions of the mucus membranes in the nasal cavity.
6 Explain how the airways in the respiratory system are similar to the branches on a tree.
7 Compare and contrast the structure of the trachea, bronchi and bronchioles.

4.2 MECHANICS OF BREATHING

For the efficient exchange of gases between the blood and the air in the alveoli, the air in the lungs must continually change. The process by which air is moved into and out of the lungs is called **ventilation**, or breathing. Air flows from places of higher pressure to places of lower pressure; therefore, air flows into and out of the lungs due to differences in air pressure.

Inspiration

The process of taking air into the lungs is called **inspiration**, or inhalation. For air to flow into the lungs, the pressure of air in the lungs must be less than the atmospheric pressure outside the body. Decreasing the pressure of air in the lungs is achieved by increasing the volume of the lungs. To increase the volume of the lungs, the diaphragm and external intercostal muscles contract. The diaphragm becomes flatter and the rib cage moves upwards and outwards, increasing the volume of the chest cavity. As the pleura adheres to the internal wall of the chest cavity, the lungs expand with the expanding chest cavity. Increased lung volume means that the air pressure inside the lungs is slightly lower than the pressure outside. Air flows in through the nose and trachea until the pressure becomes equal (Figure 4.5a). During normal, quiet breathing, the diaphragm is mainly responsible for the changes in chest volume. Movements of the rib cage become more important during heavier breathing.

Modelling breathing
Breathing animation
Watch an animation of breathing.

Expiration

Breathing out is called **expiration**, or exhalation. It occurs in the opposite way to inspiration. The diaphragm and external intercostal muscles relax, so the diaphragm bulges more into the chest cavity and the rib cage moves downwards. This reduces the volume of the chest cavity and that of the lungs. Air pressure in the lungs is now greater than pressure outside the body. Air flows out through the trachea and nose until the pressures are equal (Figure 4.5b).

When a person is breathing quietly at rest, expiration is a passive process, involving relaxation of the muscles that have contracted during inspiration. Heavier breathing involves more forceful expiration, and the intercostal muscles contract to actively lower the rib cage. The same sort of contraction occurs when forcibly exhaling, as when blowing up a balloon.

Key concept

The diaphragm and intercostal muscles work together to change the volume – and therefore pressure – of the lungs, resulting in airflow into or out of the lungs.

Activity 4.2
Investigating breathing

9780170449090

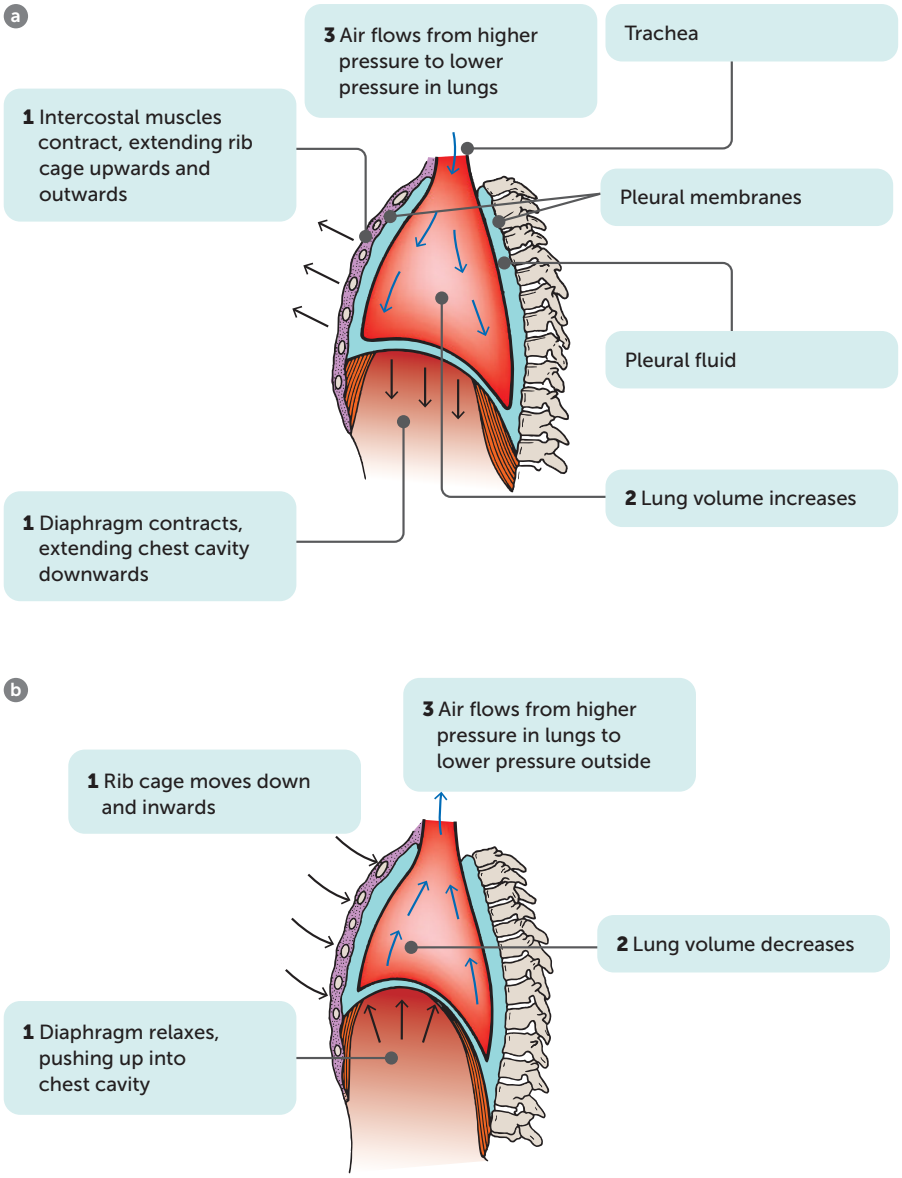


FIGURE 4.5
a Sequence of events that occur during inspiration;
b Sequence of events that occur during expiration

Questions 4.2

RECALL KNOWLEDGE

1 Describe what happens to the pressure of air in the lungs during inspiration.
2 Complete the following sentence: Air flows from areas of _____ pressure to areas of _____ pressure.
3 Use a flow chart to list the steps that occur during inspiration.

APPLY KNOWLEDGE

4 Lou Gehrig's disease, also called motor neurone disease, can cause paralysis of the diaphragm. Predict the effects of this on the body, justifying your answer.

9780170449090

4.3 GAS EXCHANGE

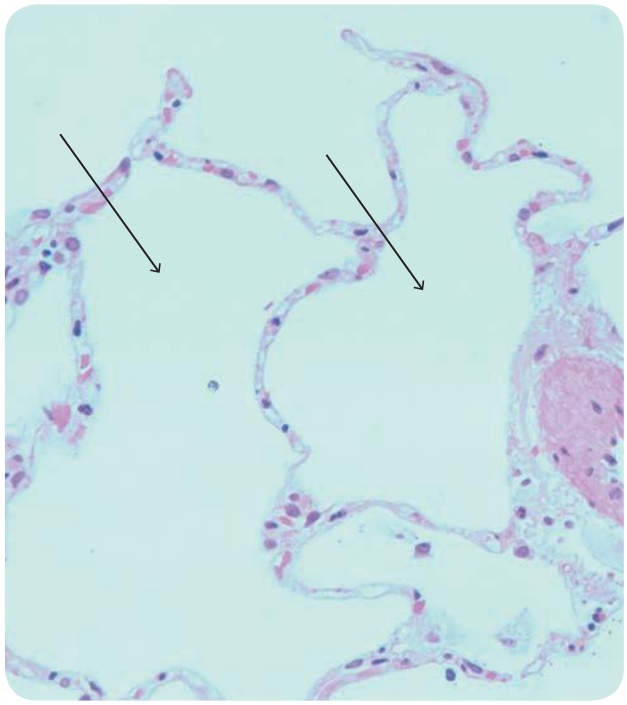
Structure of the lungs and gas exchange

The lungs are well suited to their gas exchange function, for the following reasons:

- The alveoli give the lungs a huge internal surface area, so that large amounts of gases can be exchanged in a relatively short time. Estimates of the number of alveoli in the lungs vary considerably, but there are hundreds of millions of them; they probably have a total surface area of 50–80 m² – about one-third the area of a tennis court.
- Each alveolus is well supplied with blood vessels, so that as much blood as possible is close to the air in the alveolus. The continuous flow of blood helps to maintain a difference in concentrations of oxygen and carbon dioxide between the blood and the air in the lungs.
- The membrane that forms the wall of the alveolus is very thin, so that gas molecules do not have far to travel when moving into or out of the blood. The wall has only one layer of cells and is only 1 micrometre (1 µm, or 1/1000 of a millimetre) thick.

The lungs are positioned deep inside the body to prevent excessive evaporation of the fluid that covers the respiratory surfaces. It is important that the membrane of the alveolus be covered by a thin layer of moisture because gases can diffuse into and out of the blood only when they are dissolved in fluid.

The lung volume can be changed by movements of the respiratory muscles, so that air is made to flow into and out of the lungs. Constant changing of the air in the alveoli helps to ensure that there is always a concentration gradient of oxygen and carbon dioxide between the air and the blood.



Medical Images/Carolina Biological

FIGURE 4.6 Photomicrograph of a section of lung tissue. The areas indicated by arrows are the air spaces inside the alveoli

Key concept

The structure of the lungs allows efficient gas exchange between the air and blood.

The process of gas exchange

The blood in the capillaries surrounding the alveoli is brought to the lungs by the **pulmonary arteries**. This blood has been through the capillaries of the body, where much of the oxygen has been taken up by the body cells. Therefore, the blood that comes into the capillaries around the alveoli has a low concentration of oxygen – lower than the concentration in the air in the alveolus. Oxygen dissolves in the moisture on the inside of the alveolus and diffuses through the membrane, the walls of the capillaries and into the blood.

The blood arriving at the capillaries of the alveoli has come from the body circulation where it has picked up carbon dioxide produced by respiration in the cells. The concentration of carbon dioxide in the alveolar capillaries is higher than the concentration in the air in the alveolus. Therefore, carbon dioxide diffuses out of the blood and into the air in the alveolus.

9780170449090

Gas exchange
This website includes an animation of gas exchange.

In summary, expired air contains less oxygen and more carbon dioxide than inspired air.

TABLE 4.1 Oxygen and carbon dioxide concentrations in inspired and expired air

	INSPIRED AIR (%)	EXPIRED AIR (%)
Oxygen	20.95	15.80
Carbon dioxide	0.04	4.30

Note: The other 79% of the inspired air is made up mainly of nitrogen, with varying amounts of water vapour.

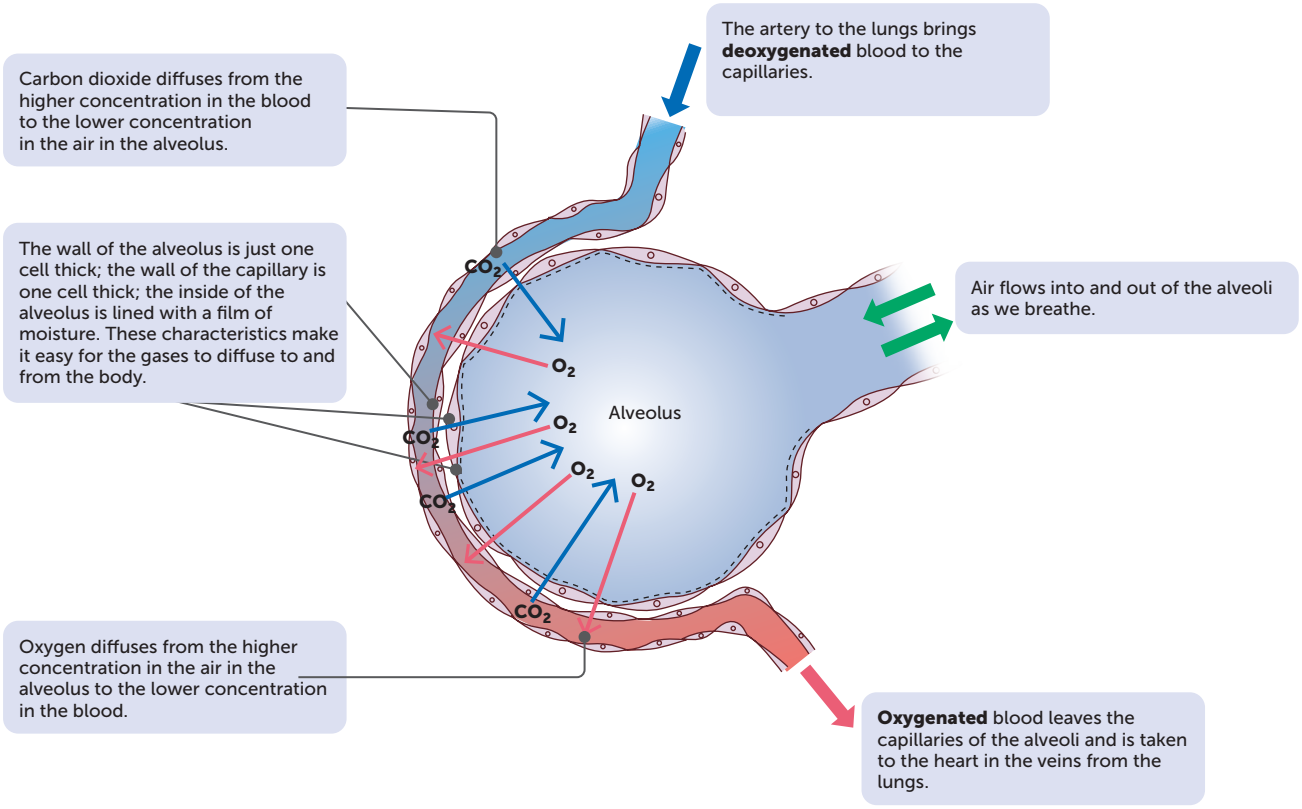


FIGURE 4.7 Gas exchange between alveolar air and blood

For diffusion of gases into and out of the blood, there must be a **concentration gradient** – that is, a difference in gas concentration between the air in the alveoli and the blood in the capillaries. The concentration gradient for oxygen and carbon dioxide is maintained by:

- the constant flow of blood through the capillaries. As the blood flowing through the capillaries around each alveolus picks up oxygen and loses carbon dioxide, it is replaced by more blood being pumped into the capillaries. This 'new' blood is low in oxygen and high in carbon dioxide, so the concentration gradient is maintained
- the movement of air into and out of the alveoli as we breathe in and out. The air that has picked up carbon dioxide from, and lost oxygen to, the blood is replaced by 'new' air with each breath. The 'new' air is low in carbon dioxide and high in oxygen.

Key concept

There is a net diffusion of oxygen into the blood and carbon dioxide out of the blood due to the concentration gradients between the air and blood.

Questions 4.3

RECALL KNOWLEDGE

1 How thick is the wall of the alveolus?
2 Describe the movement of oxygen between the air and the blood.
3 Explain why the concentration of carbon dioxide in the blood in the capillaries surrounding the alveoli is higher than in the air within the alveoli.
4 Explain why it is important that there is a constant flow of blood through the capillaries surrounding the alveoli.
5 Explain how the pleural fluid plays an important role in gas exchange.

APPLY KNOWLEDGE

6 People with cystic fibrosis produce an excessive amount of thick mucus, primarily in the lungs and digestive system. Explain how this would affect the respiratory system, and predict the symptoms exhibited.

4.4 SOME EFFECTS OF LIFESTYLE AND ENVIRONMENT ON GAS EXCHANGE

How we live, and the environments in which we work and spend our leisure time, profoundly affect the efficiency of the respiratory surfaces.

Emphysema

Emphysema is a disease usually caused by long-term exposure to irritating particles in the air taken into the lungs. While no one can avoid inhaling particles, as there are always particles of matter in the air, some people are exposed to excessively high levels. Smokers constantly inhale irritants in tobacco smoke. People who work in situations where a lot of dust is produced are also at risk. There is also a greater risk of emphysema in cities with continually high air pollution.

The irritating particles cause damage to the alveoli. They lose their elasticity, are often replaced with fibrous tissue, and may break down, reducing the internal surface area of the lung (Figures 4.8 and 4.9). Because of the loss of elasticity of the lung tissue, the lungs are constantly inflated, and breathing out no longer occurs passively but requires voluntary effort. Thus, the emphysema sufferer has two problems – inadequate surface area for gas exchange, and difficulty in ventilating the lungs. Emphysema cannot be cured, and once lung damage begins, the progression of the disease cannot be stopped.

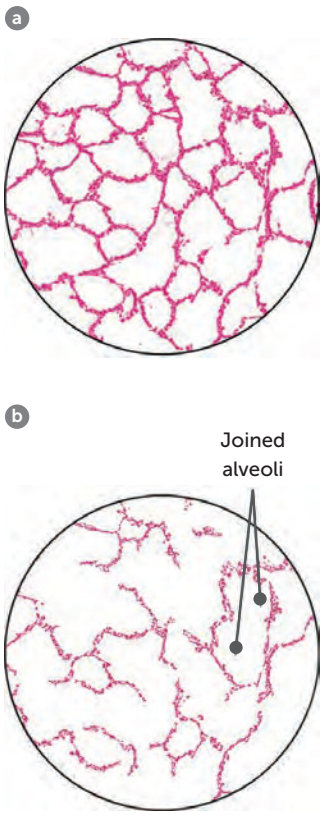


FIGURE 4.8
a Photomicrograph of a section of lung tissue showing normal alveoli; **b** Photomicrograph of a section of lung tissue from a patient with emphysema showing how alveolar walls have broken down, forming larger, fewer alveoli with reduced total surface area

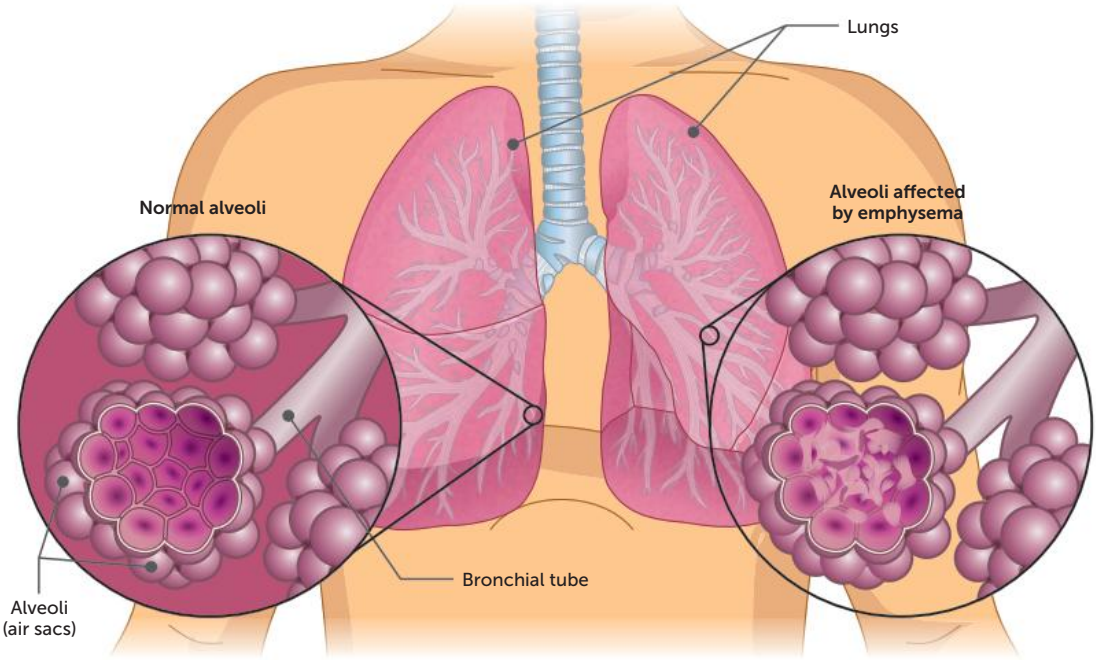
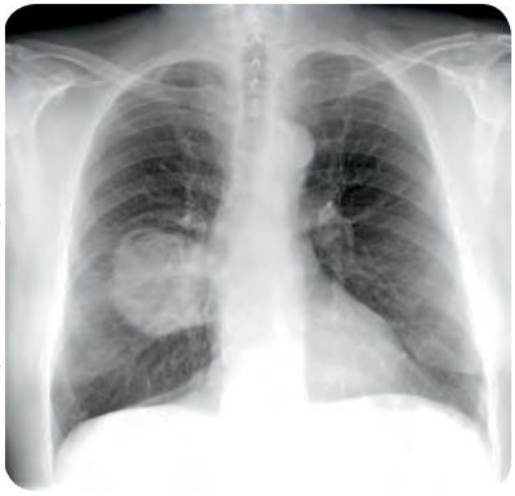


FIGURE 4.9 Normal alveoli compared to alveoli affected by emphysema



Science Photo Library/Du Cane Medical Imaging Ltd.

FIGURE 4.10 X-ray of a patient with a lung cancer tumour

Lung cancer

Lung cancer is similar to most other cancers in that it involves the development of a mass of cells that divides in an uncontrolled way – that is, a tumour. Evidence shows clear links between lung cancer and exposure to asbestos fibres and other pollutants. However, tobacco smoking poses by far the greatest risk for lung cancer. This risk increases if smoking is combined with other risk factors. For example, people who have worked with asbestos and who also smoke have a 20–90 times greater risk of contracting lung cancer than similar workers who do not smoke. Some chemical substances seem to initiate cancerous growths; others seem to promote the growth of the tumour. Tobacco smoke contains both initiators and promoters of lung cancer.

The most common form of lung cancer begins in the walls of the air passages, usually the bronchi. Inhaled smoke particles constantly irritate the mucous membrane that lines the air passages. This results in excessive production of mucus. Cells at the base of the membrane begin to divide more rapidly and the accumulating mucus cannot be removed. This results in 'smoker's cough'. The trapped mucus causes rupture of the alveoli. Emphysema has then developed. Ultimately, a cancerous growth develops in an air passage and may spread to other parts of the body.

Lung infections

Pneumonia is an infection of the lungs caused by bacteria, viruses, fungi or other organisms. The inflammation resulting from the infection causes secretion of fluid and mucus into the alveoli, thus reducing the amount of air that they can contain. The surface area available for exchange of gases is also reduced, and breathing difficulty is a symptom of many types of pneumonia.

9780170449090

Tuberculosis (TB) is an infection, usually of the lung, by the bacterium *Mycobacterium tuberculosis*. Tuberculosis, along with HIV/AIDS and malaria, is one of the top three infectious diseases causing death in the world. Fortunately, in Australia there is a very low incidence of tuberculosis. There are only about 1000 cases per year, with most of them in people born overseas.

Most lung infections, such as tuberculosis and pneumonia, are spread by droplets. When infected people cough, sneeze or spit, tiny droplets of moisture containing the bacteria, viruses or fungi may be inhaled by others, causing the spread of the infection. Good hygiene practices, such as coughing and sneezing into a handkerchief and not spitting, help to reduce the spread of lung infections.

Asthma

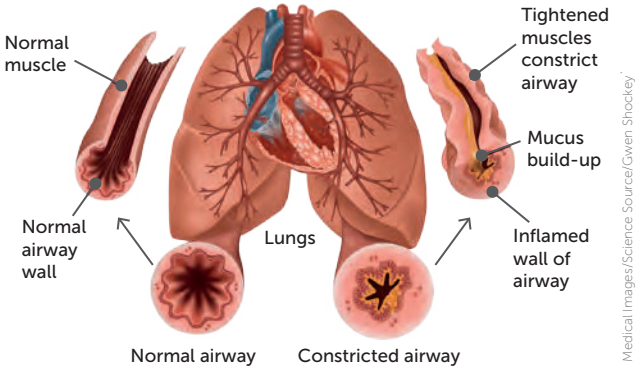
Asthma is a medical condition that causes difficulty breathing due to a narrowing of the airways. This occurs due to:

- the smooth muscles contracting, narrowing the airway
- inflammation causing the lining of the airways to thicken, narrowing its diameter
- mucus filling the airway, narrowing the tube.

Asthma can be an allergic response or non-allergic. Triggers for an asthma flare-up include respiratory infections, cigarette smoke, and allergens such as dust, mites, moulds, pollens and animals. Other triggers include exercise, cold weather, smoke, some medications, stress and emotions. Some substances in food may also trigger an asthma attack.

During such an attack, the muscles that surround the bronchioles go into spasm, which is sudden involuntary contractions. This causes a narrowing of the air passages and, therefore, difficulty in breathing.

Usually the irritation of the membranes lining the air passages also causes secretion of excessive mucus, which also restricts the movement of air. The reduced volume of air going into and out of the lungs means that the exchange of gases is impaired, and the blood does not carry the usual amount of oxygen.



Medical Images/Science Source/Gwen Shockey

FIGURE 4.11
During an asthma attack, the smooth muscles cause the bronchioles to narrow

Key concept

Factors in an individual's lifestyle can affect their respiratory system, causing disorders and diseases such as emphysema, lung cancer, lung infections and asthma.

Questions 4.4

RECALL KNOWLEDGE

1. Name the process that results in the alveoli breaking down and reducing the surface area.
2. Describe the changes that occur in the bronchioles during an asthma attack.
3. List the most common causes of lung cancer.

APPLY KNOWLEDGE

4. Explain why pneumonia results in the patient feeling very tired.
5. Construct a table to compare the cause, symptoms and treatment of emphysema, lung cancer, lung infections and asthma.

CHAPTER 4 ACTIVITIES

ACTIVITY 4.1 Examining the structure of the lungs

This activity may be done as a demonstration by your teacher.

What to do

Examine a set of sheep or pig lungs.

1. Identify the structures that can be seen, such as:
   - **a** the lungs themselves, divided into a number of lobes
   - **b** the trachea with its rings of cartilage; examine the rings to see whether they form a complete circle
   - **c** the two bronchi that branch from the trachea
   - **d** the thin transparent membrane that covers the lungs.
2. Squeeze the lungs between your thumb and a finger. Describe what you feel.
3. Cut off a piece of lung and place it in a beaker of water. Does it float? What does this tell you about the lung?
4. Cut open the trachea and observe the interior. Record your observations.
5. Continue the cut in the trachea down through one of the bronchi, then through a secondary bronchus. Keep cutting until the air tubes become too small to see. Do the secondary bronchi have rings of cartilage? As you go along the air tubes from large to small, where do the cartilage rings stop?

ACTIVITY 4.2 Investigating breathing

This task will allow you to investigate a factor that affects the rate of breathing.

What to do

1. Brainstorm all the factors that could affect the rate of breathing.
2. Choose one of these factors to investigate.
3. Develop a hypothesis stating the expected trend between this factor and the rate of breathing.
4. Design an experiment to test your hypothesis.
   - **a** How will you change your independent variable (the factor that you are testing)?
   - **b** How will you measure your dependent variable (the breathing rate)?
   - **c** How will you control all the other factors?
   - **d** How many trials will you conduct?
5. Under your teacher's direction, conduct your investigation.
6. Collate your data in a table and calculate the average breathing rate for each variation of the factor you are testing.
7. Use your results to construct a graph.
   - **a** What type of graph should you use?
   - **b** Which variable is on the x-axis? Which variable is on the y-axis?
   - **c** What scale will you use?
   - **d** Draw the line of best fit for the data.

Studying your results

1. What trend does your data show? That is, what is the relationship between the independent and dependent variables?
2. Discuss the reliability of your data. That is, how consistent are your results for the trials?
3. Discuss the accuracy of your results. That is, how did you ensure that your measurements were correct?
4. Discuss the validity of your method. That is, how well did you control your variables? How well did you test the hypothesis?
5. Discuss whether your results support your hypothesis.

9780170449090

CHAPTER 4 SUMMARY

- Air enters, and leaves, the body through the nose, mouth, pharynx and larynx.
- The air is warmed and humidified prior to entering the lungs.
- The trachea takes the air to and from the lungs. It contains C-shaped cartilage rings that hold the structure open.
- The trachea divides, forming the primary, then secondary and tertiary bronchi. As the bronchi get smaller, there are fewer cartilage rings and more smooth muscle and elastin.
- The bronchioles are finer than the bronchi and do not contain cartilage.
- The bronchioles end in air sacs called alveoli, which are the site of gas exchange.
- As the air moves through the trachea and bronchi, mucus and cilia work together to trap and remove foreign particles.
- The lungs are covered in a membrane that extends to the inside of the chest. Pleural fluid is contained between the two layers of the membrane and allows the lungs to move while still holding them to the side of the chest.
- Air will flow from areas of high pressure to areas of low pressure.
- During inspiration the diaphragm and intercostal muscles contract, increasing the volume of the lungs and decreasing the pressure. This causes air to flow into the lungs.
- During expiration the diaphragm and intercostal muscles relax, decreasing the volume of the lungs and increasing the pressure. This causes air to flow out of the lungs.
- Gas exchange occurs at the alveoli.
- Alveoli allow efficient gas exchange as they have a very large surface area, have a good blood supply, are very thin, are covered by a thin layer of moisture and have a constant movement of air.
- The concentration of oxygen in the blood is lower than in the alveoli, resulting in a net diffusion of oxygen into the blood.
- The concentration of carbon dioxide is higher in the blood than in the air; therefore, there is a net diffusion of carbon dioxide out of the blood.
- Our lifestyle may affect the respiratory system:
  - Emphysema is caused by long-term exposure to irritants such as tobacco smoke and air pollutants. It causes the alveoli to break down, reducing the surface area for gas exchange.
  - Lung cancer is the uncontrolled division of cells. This may cause a mass, excess mucus and the rupture of alveoli. The presence of lung cancer is linked to smoking and exposure to asbestos and other pollutants.
- Infections such as pneumonia and tuberculosis can cause inflammation, resulting in secretions in the alveoli. This reduces the surface area for gas exchange as well as the volume of air in the lungs.
- Asthma causes a narrowing of the airways due to contraction of the smooth muscles, inflammation of the lining, and mucus filling the airways. These combine to cause breathing difficulties.

9780170449090

CHAPTER 4 GLOSSARY

Alveoli Air sacs in the lungs; also, the milk-secreting part of the mammary glands; singular: alveolus

Asthma An allergic condition that causes narrowing of the airways and difficulty breathing

Bronchiole A very small air tube in the lung

Concentration gradient A difference in concentration of a solution, often between the inside and outside of a cell; also called diffusion gradient

Emphysema A disease of the lungs that damages the alveoli; caused by long-term exposure to irritants

Epiglottis A cartilage flap at the base of the pharynx that covers the trachea during swallowing

Expiration Breathing out; exhalation

Inspiration Breathing in; inhalation

Larynx The structure at the top of the trachea that contains the vocal cords

Lung cancer Growth of a tumour in the lungs

Oesophagus The tube that carries food from the throat to the stomach

Pharynx The throat; the pharynx joins the mouth cavity to the oesophagus and larynx

Pleura A membrane covering the surface of the lungs

Pleural fluid A thin layer of fluid within the pleura that allows the lungs to move during breathing

Pneumonia A lung infection that causes fluid and mucus to build up in the alveoli, causing difficulty breathing

Primary bronchi The first branching from the trachea, entering the left and right lungs

Pulmonary arteries The artery that takes blood from the heart to the lungs

Respiratory system The system specialised to facilitate the intake of oxygen and the removal of carbon dioxide

Secondary bronchi The division of the primary bronchi, taking air into each lobe of the lungs

Terminal bronchioles The end of the bronchioles before they form alveoli

Tertiary bronchi The division of the secondary bronchi

Trachea The tube that takes air from the throat to the lungs; the windpipe

Tuberculosis (TB) A lung infection caused by the bacterium *Mycobacterium tuberculosis*

Ventilation The process of inhalation and exhalation; breathing

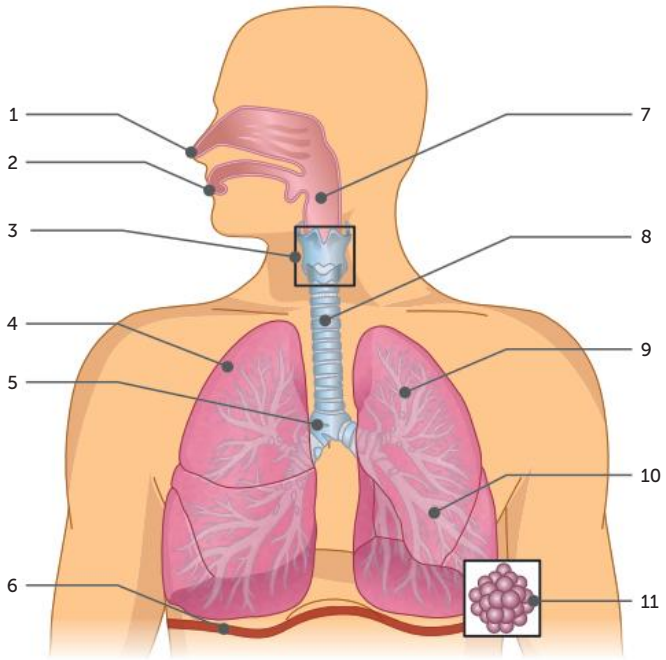
Vocal cord Membrane in the larynx that vibrates, producing sounds

9780170449090

CHAPTER 4 REVIEW QUESTIONS

Recall

1 Label the parts of the respiratory system.



2 a Draw a diagram showing inspiration. As labels for your diagram, list the sequence of events that occur in inspiration.

b Draw a diagram showing expiration. As labels for your diagram, list the sequence of events that occur in expiration.

3 List the characteristics of the lungs that make them well suited for gas exchange.

4 Describe precautions that you can take to reduce your risk of developing emphysema or lung cancer.

Explain

5 Explain why it is important that there is cartilage in the trachea and bronchi

6 a Why is a concentration gradient important for the exchange of gases?

b Why is it that, in the lungs, oxygen diffuses into and carbon dioxide out of the blood, whereas in other body tissues oxygen diffuses out of and carbon dioxide into the blood?

c Explain how a concentration gradient for oxygen and carbon dioxide is maintained between the blood and the air in the alveoli.

7 Explain why the pressure in the lungs decreases during inspiration.

8 Explain why asthma is such a serious condition.

9780170449090

Apply

9 Compare and contrast bronchi and bronchioles.

10 To be effective, any surface where materials are taken into the body, or passed out of the body, must have a very large surface area. For the lungs, explain how a large surface area is achieved.

11 List five occupations in which people could be at risk of contracting emphysema. What precautions could be taken to reduce the risk of workers contracting the disease?

12 Students measured the breathing rate and depth of breathing of a girl before and after exercise. Their results are shown in the following table.

	BREATHS PER MINUTE	VOLUME OF AIR PER BREATH (cm^3)
At rest	19	460
After running	38	1075

a Calculate the total volume of air that the girl breathed in 1 minute before and after exercise.

b What is the reason for the increase in rate and depth of breathing after exercise?

c Describe the changes that would occur in the body to bring breathing back to the normal resting level after exercise.

13 Describe the types of lung damage that smoking can cause.

Extend

14 If air enters the chest cavity through a puncture wound to the chest wall, the lung may collapse. As the collapsed lung is no longer attached to the chest wall, air cannot be made to move into and out of the lung. However, a person with a collapsed lung can function fairly normally.

a Explain how it would be possible for such a person to function in a fairly normal way.

b Would there be any activities that such a person would not be able to perform?

15 The ability to voluntarily control breathing is important when speaking, but it is also important when eating or drinking. Explain why this is so.

16 In expired air resuscitation (mouth-to-mouth resuscitation), air from the rescuer's lungs is blown into the patient's lungs. How is expired air able to keep the patient alive?

9780170449090

5 THE CIRCULATORY SYSTEM TRANSPORTS MATERIALS THROUGHOUT THE BODY

UNIT 1 CONTENT

SCIENCE INQUIRY SKILLS

- conduct investigations, including monitoring body functions; use microscopy techniques; and perform real or virtual dissection, safely, competently and methodically for the collection of valid and reliable data
- interpret a range of scientific and media texts, and evaluate processes, claims and conclusions by considering the quality of available evidence; and use reasoning to construct scientific arguments

SCIENCE AS A HUMAN ENDEAVOUR

- blood transfusions rely on determining blood groups and can be used to treat many different diseases and conditions

SCIENCE UNDERSTANDING

Circulatory system

- the transport of materials within the internal environment for exchange with cells is facilitated by the structure and function of the circulatory system at the cell, tissue and organ levels
- the components of blood facilitate the transport of different materials around the body (plasma and erythrocytes), play a role in the clotting of blood (platelets) and the protection of the body (leucocytes)
- the lymphatic system functions to return tissue fluid to the circulatory system and to assist in protecting the body from disease

School Curriculum and Standards Authority, Government of Western Australia

9780170449090

The body's main internal transport system is the **circulatory system**. It is the link between the cells inside the body, which have certain requirements, and the environment outside the body, which supplies those requirements.

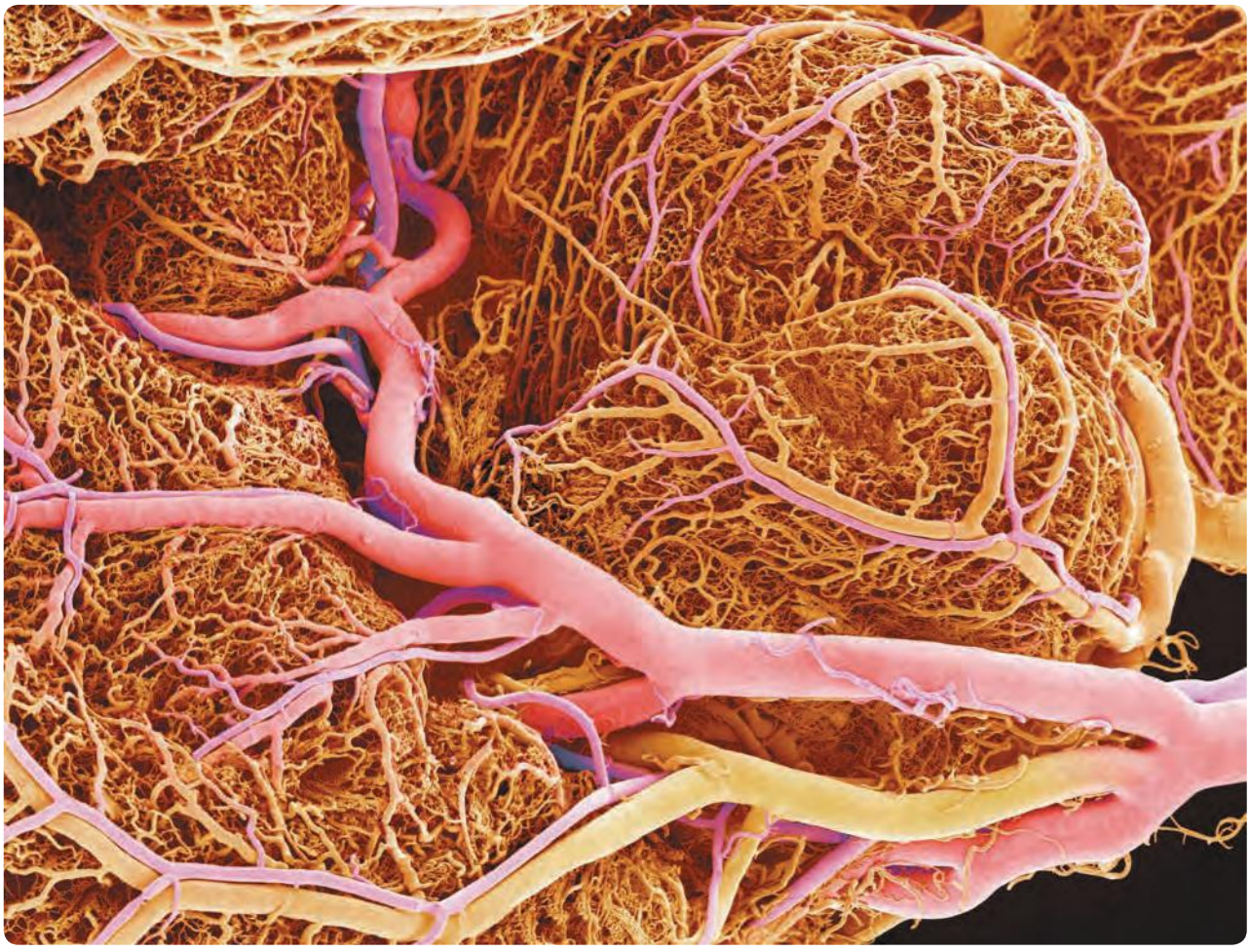
Special organs are needed to extract these requirements from the environment. The digestive system absorbs nutrients and the respiratory system absorbs oxygen.

Other organs pass waste from the body to the environment. The respiratory system excretes carbon dioxide, and the kidneys excrete other wastes.

Blood is the transport link between the cells of all the body systems. It is also very important in maintaining the constant internal environment of the body. Some of the more important functions of blood are:

- transporting oxygen and nutrients to all cells of the body
- transporting carbon dioxide and other waste products away from the cells
- transporting chemical messengers, called hormones, to the cells
- maintaining the pH of body fluids
- distributing heat and maintaining body temperature
- maintaining water content and ion concentration of the body fluids
- protecting against disease-causing micro-organisms
- clotting when vessels are damaged, thus preventing blood loss.

This chapter looks at how the structure of the blood, heart and blood vessels enables the functions listed above to be achieved.



Alamy Stock Photo

FIGURE 5.1 Coloured scanning electron micrograph (SEM) of blood vessels in the small intestine. Substances are taken to and removed from cells via the blood, which circulates through arteries, capillaries and veins

9780170449090

5.1 BLOOD AS A TRANSPORT MEDIUM

An average female adult has approximately 4–5 L of blood, whereas an adult male has about 5–6 L.

The structure of blood

Blood is composed of:

- plasma: the liquid part, making up approximately 55% of the blood volume
- **formed elements**: the non-liquid part, making up 45% of the blood volume and consisting of erythrocytes (red blood cells), leucocytes (white blood cells) and thrombocytes (platelets).

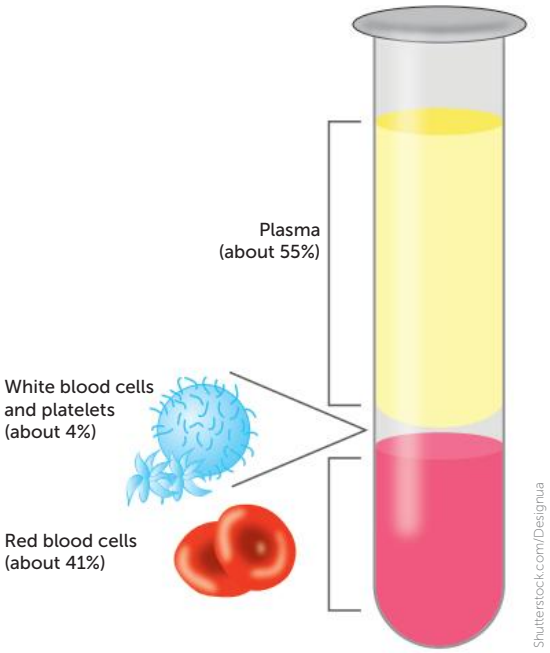


FIGURE 5.2 The composition of blood

Plasma

Plasma is a mixture of water with dissolved substances such as sugar and salts. The function of plasma is to transport the components of blood, including cells, nutrients, wastes, hormones, proteins and antibodies, throughout the body.

Erythrocytes

Erythrocytes, or **red blood cells**, are the most abundant cells in the blood and account for approximately 40–45% of its volume. This percentage is known as the **haematocrit**. The cells are a **biconcave** shape – flattened in the middle on both sides. Red blood cells do not contain a nucleus, which increases their flexibility and, hence, their ability to move through blood vessels. However, the lack of a nucleus also limits their life span to only 120 days on average. The function of the erythrocytes is to transport oxygen from the lungs to the cells throughout the body.

Leucocytes

Leucocytes, or **white blood cells**, play an important role in protecting the body from infection. While they make up only 1% of the blood, white blood cells are larger than red blood cells.

There are a number of different leucocytes, each with its own structure and function.

- Neutrophils are the most common type of white blood cells. They contain enzymes to digest pathogens.

9780170449090

- Monocytes form other cells, including macrophages that engulf pathogens and aged or damaged cells by phagocytosis.
- Lymphocytes are involved in the immune response; cell-mediated immunity uses T-lymphocytes, and antibody-mediated immunity uses B-lymphocytes.
- Basophils are responsible for allergic reactions, producing heparin and histamine to defend the body against parasites and bacteria.
- Eosinophils also lead to inflammatory responses; they respond to larger parasites such as worms.

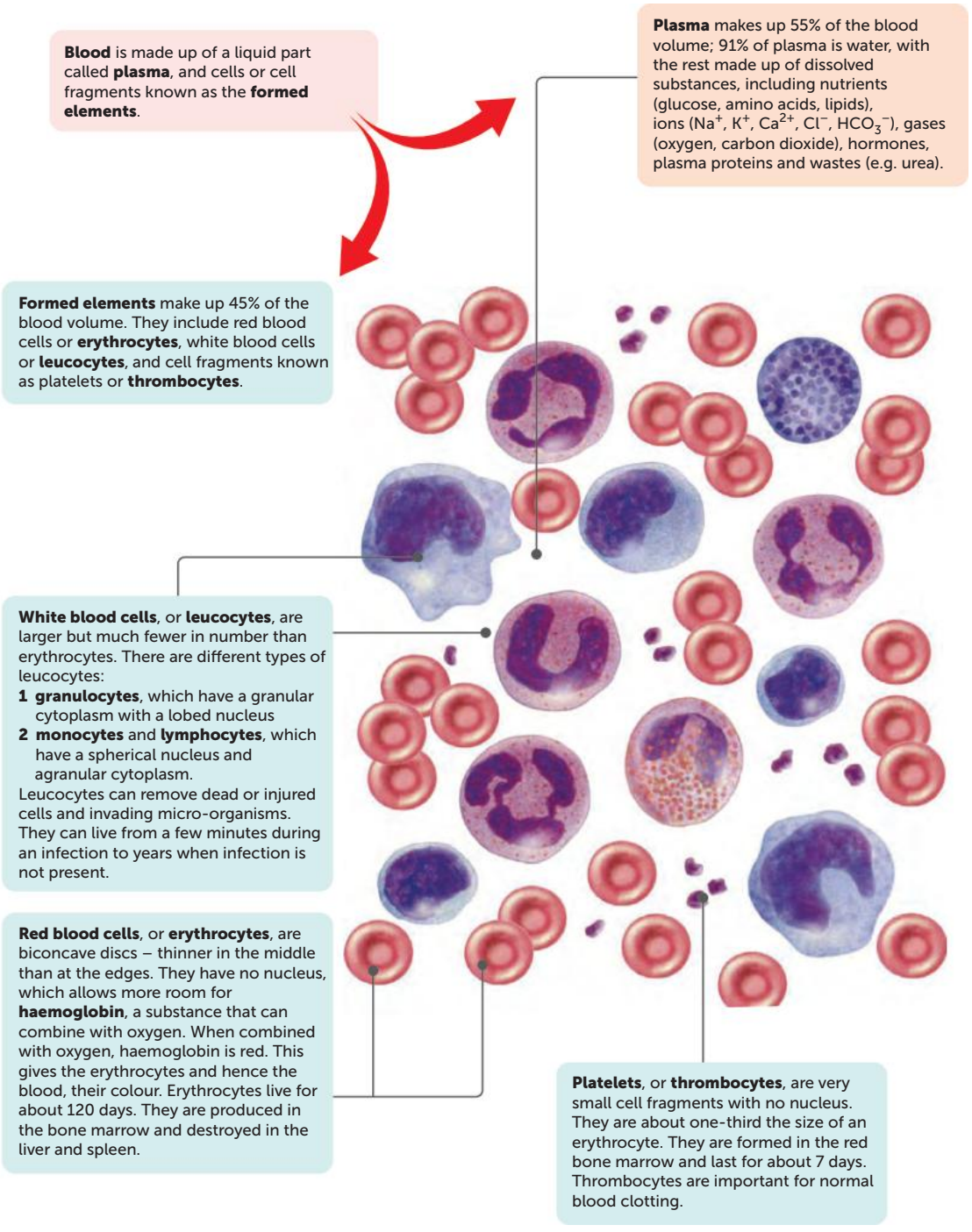


FIGURE 5.3 Blood is made up of plasma and formed elements

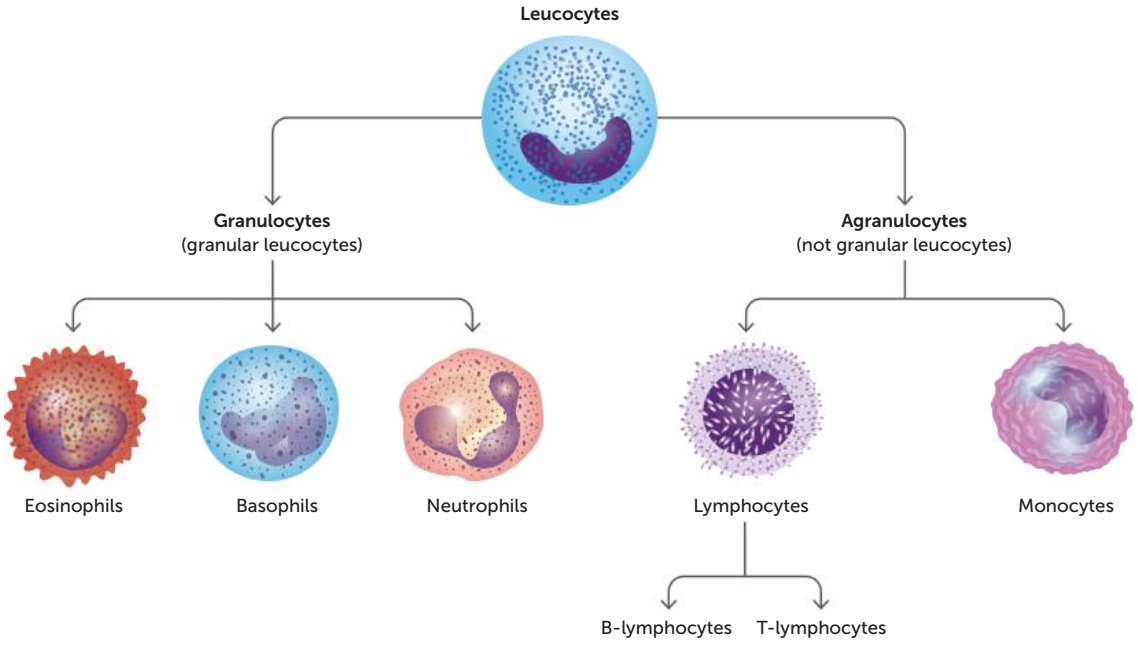


FIGURE 5.4 Types of leucocytes

Thrombocytes

Thrombocytes, or **platelets**, are small fragments of cells. When a blood vessel is injured, the platelets adhere to the lining and form a scaffold for the coagulation of the blood to form a clot.

Red Cross
Learn about the components of blood.

Key concept

Blood is made up of plasma, erythrocytes, leucocytes and thrombocytes; each component performs an important role in the body.

Activity 5.1
Comparing blood cells

Transport of oxygen

Oxygen is not very soluble in water, so only about 3% of oxygen is carried in solution in the blood plasma. The other 97% is carried in combination with haemoglobin molecules, which are found only in red blood cells. **Haemoglobin** is able to combine with oxygen to form a compound called **oxyhaemoglobin**. The combination of oxygen and haemoglobin is said to be a *loose* one, because oxyhaemoglobin can easily break down to release the oxygen:

$$\underset{\text{Haemoglobin}}{Hb} + \underset{\text{Oxygen}}{O_2} \rightleftharpoons \underset{\text{Oxyhaemoglobin}}{HbO_2}$$

The presence of haemoglobin in the red blood cells increases the oxygen-carrying capacity of the blood by about 60 or 70 times.

Oxygen combines with haemoglobin when the oxygen concentration is relatively high. This occurs in the capillaries in the lungs, where oxygen diffuses into the blood from the air in the alveoli. Oxyhaemoglobin breaks down to haemoglobin and oxygen when the concentration of oxygen is relatively low. As the cells of the body are continually using oxygen, the tissue fluid around the cells has a relatively low oxygen concentration. Therefore, when the red blood cells flow through the capillaries between the body cells, they give up their oxygen, which diffuses into the tissue fluid and then into the cells.

Oxygenated blood is blood with a high proportion of oxyhaemoglobin. Oxyhaemoglobin is bright red in colour, so the blood in the arteries (except for the pulmonary arteries taking blood to the lungs) is bright red. Haemoglobin is dark red or purplish in colour. The **deoxygenated blood** in the veins (except for the pulmonary veins from the lungs) is therefore dark red.

Red blood cells are well suited to their function of oxygen transport because they:

- contain haemoglobin, which is able to combine with oxygen
- have no nucleus, so there is more room for haemoglobin molecules
- are shaped like biconcave discs – the biconcave centre increases the surface area for oxygen exchange and the thicker edges give a large volume that allows room for the haemoglobin molecules.

Key concept

The majority of oxygen is transported on erythrocytes as oxyhaemoglobin.

Activity 5.2
Investigating blood flow during exercise

Transport of carbon dioxide

Carbon dioxide is carried in the blood in a number of ways. Approximately 7–8% is dissolved in the plasma and carried in solution. Another 22% or so combines with the globin part of the haemoglobin molecule to form a compound called **carbaminohaemoglobin**. The remainder, about 70%, is carried in the plasma as bicarbonate ions, HCO_3^-.

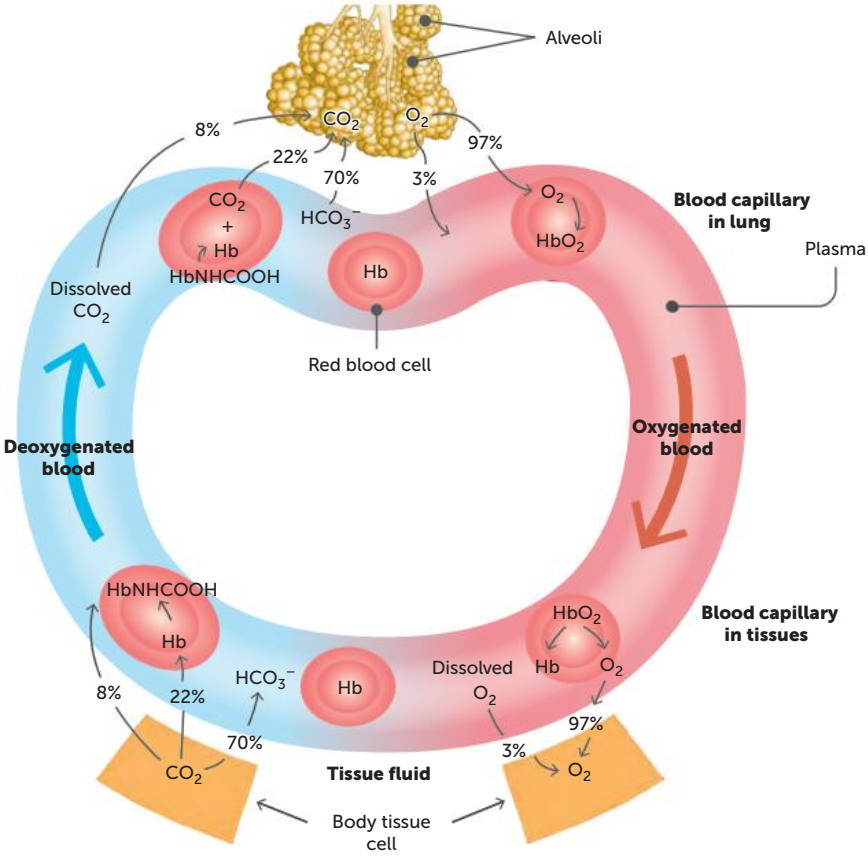


FIGURE 5.5 The transport of oxygen and carbon dioxide

TABLE 5.1 Proportion of oxygen and carbon dioxide transported in the blood in different ways

OXYGEN	CARBON DIOXIDE
3% dissolved in plasma	8% dissolved in plasma
97% as oxyhaemoglobin	22% as carbaminohaemoglobin
	70% as bicarbonate ions

As the blood is flowing through the capillaries between the body cells, carbon dioxide diffuses into the plasma due to the difference in carbon dioxide concentration. Some carbon dioxide dissolves in the plasma, some combines with haemoglobin, but most reacts with water to form carbonic acid (H_2CO_3). Carbonic acid then ionises into hydrogen ions and bicarbonate ions:

$$\underset{\text{Carbon dioxide}}{CO_2} + \underset{\text{Water}}{H_2O} \rightleftharpoons \underset{\text{Carbonic acid}}{H_2CO_3} \rightleftharpoons \underset{\text{Hydrogen ions}}{H^+} + \underset{\text{Bicarbonate ions}}{HCO_3^-}$$

The air sacs of the lungs, the **alveoli**, are surrounded by a dense network of capillaries (see Figure 5.1). Here, the carbon dioxide dissolved in the plasma diffuses out of the blood into the air in the alveolus. The carbaminohaemoglobin breaks down, and the carbon dioxide molecules released also diffuse into the alveolus. Hydrogen ions and bicarbonate ions recombine to form carbonic acid, which then breaks down under enzyme action into water and carbon dioxide. This carbon dioxide also diffuses into the alveolus (see Figure 5.5 and Table 5.1).

Key concept

Carbon dioxide can be transported by dissolving in plasma (7–8%), as carbaminohaemoglobin (22%) or as bicarbonate ions (70%) in the plasma.

Transport of nutrients and waste

Nutrients and wastes (apart from carbon dioxide, which we have already discussed) are dissolved and transported in the blood plasma.

Nutrients are the essential elements and molecules that are obtained from the food we eat. Inorganic nutrients are transported as ions. Some of the important ions dissolved in the blood plasma are sodium ions (Na^+), calcium ions (Ca^{2+}), potassium ions (K^+), chloride ions (Cl^-) and iodide ions (I^-). Organic nutrients dissolved in the blood plasma include glucose, vitamins, amino acids, fatty acids and glycerol.

Wastes, or more correctly, **metabolic wastes**, are substances produced by the cells that cannot be used and would be harmful if allowed to accumulate. The most important organic wastes that are transported in solution in the blood plasma are urea, creatinine and uric acid.

Blood clotting

When an injury occurs that involves damage to blood vessels, the events that follow help to minimise blood loss from the broken vessels and prevent the entry of infecting micro-organisms.

1. *Vasoconstriction*: The muscles in the walls of the small arteries that have been injured or broken constrict immediately to reduce blood flow and, therefore, blood loss.
2. *Platelet plug*: The internal walls of blood vessels are normally very smooth, but any damage creates a rough surface to which the platelets stick. Sticking platelets attract others, and so a plug is built up at the site of the injury. This plug also helps to reduce blood loss. The platelets release substances that act as vasoconstrictors, which enhance and prolong the constriction of the damaged vessels. For many of the small tears that occur in capillaries each day, this plugging action of the platelets and constriction of the blood vessels is sufficient to stop any bleeding.
3. *Coagulation*: For more serious injuries, **blood clotting**, or **coagulation**, is necessary. The formation of a blood clot is a complex process involving a large number of chemical substances, or **clotting factors**, that are present in the blood plasma. The complex series of reactions results in the formation of threads of an insoluble protein called **fibrin**. The fibrin threads form a mesh that traps blood cells, platelets and plasma. This mesh, with its trapped material, is the **clot** or **thrombus**. The threads stick to the damaged blood vessels and hold the clot in position.

9780170449090

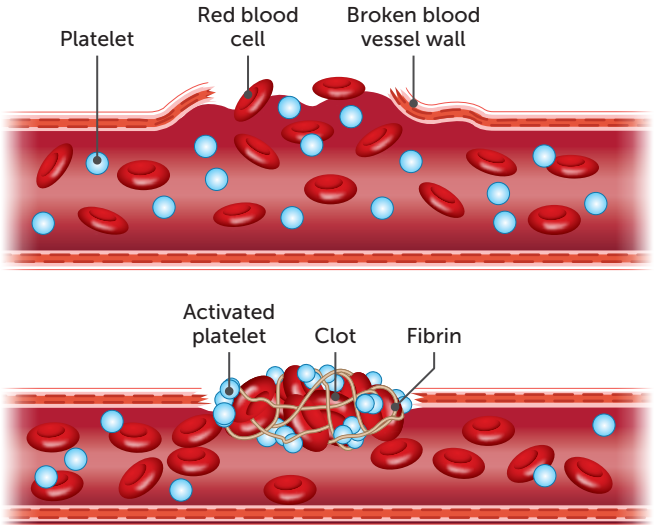
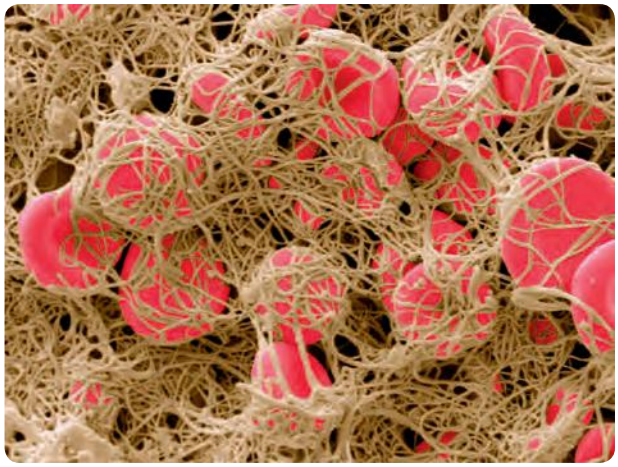


FIGURE 5.6 The process of blood clotting



Alamy Stock Photo/Science Photo Library

FIGURE 5.7 Scanning electron micrograph of a blood clot

Blood clotting
Watch an animation of blood clotting.

After the formation of the clot, a slower process known as **clot retraction** occurs. The network of threads contracts, becoming denser and stronger and pulling the edges of the damaged blood vessels together. As clot retraction occurs, a fluid known as **serum** is squeezed out. The clot then dries, forming a scab over the wound that prevents entry of infecting micro-organisms.

Questions 5.1

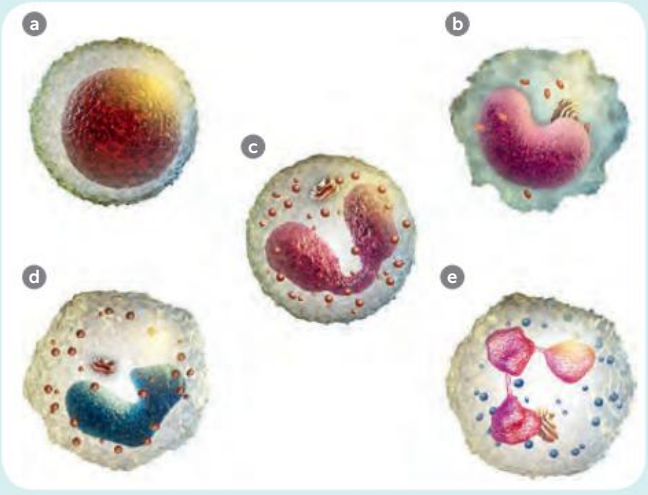
RECALL KNOWLEDGE

1 State the common name for:
   a leucocytes
   b thrombocytes
   c erythrocytes.

2 Copy and complete the table below regarding the composition of blood.

STRUCTURE OF BLOOD	PERCENTAGE BY VOLUME (%)
Plasma	
	40–45
Leucocytes	
	<1

4 Name each type of white blood cell in the diagram below.



Shutterstock.com/Andrea Danti

5 Describe how monocytes are able to protect the body.
6 Explain how oxygen is transported from the alveoli to the body cells.
7 Draw a flow chart to show the steps involved in blood clotting.

APPLY KNOWLEDGE

8 One side effect of chemotherapy is thrombocytopenia, a condition characterised by a low platelet count. Predict the symptoms that would be evident due to this condition.
9 Discuss the advantages and disadvantages of erythrocytes lacking a nucleus.

5.2 MOVING BLOOD THROUGH THE BODY

The heart

The **heart** is the pump that pushes the blood around the body. It is located between the two lungs in the mediastinum, behind and slightly to the left of the **sternum**.

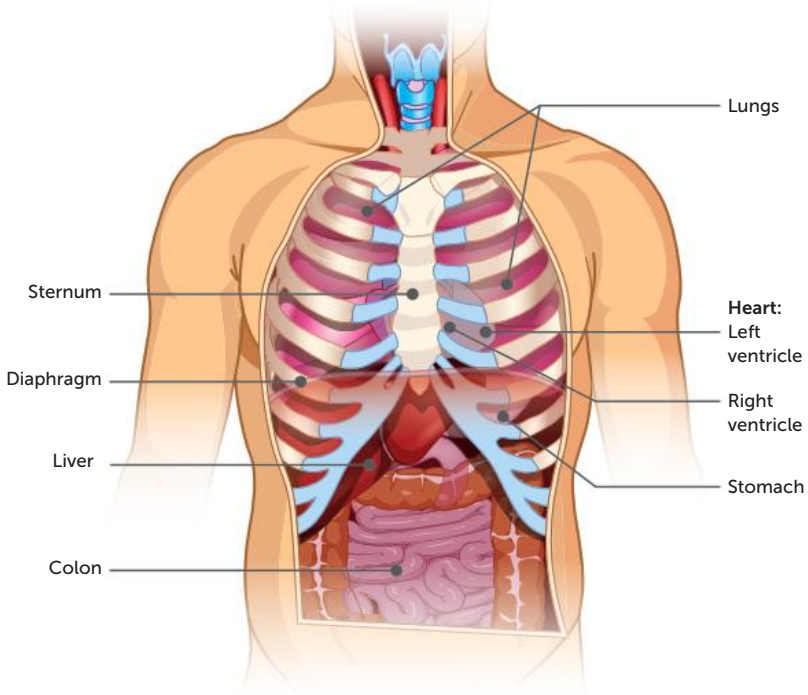


FIGURE 5.8 Location of the heart

9780170449090

The heart is a conical shape approximately 12 cm long, 9 cm at its widest point and 6 cm thick, making it about the size of an adult human fist. It is completely enclosed in a membrane called the **pericardium**. This membrane holds the heart in place, but also allows the heart to move as it beats. It also prevents the heart from overstretching. The wall of the heart itself is made up of a special type of muscle, called **cardiac muscle**.

The left and right sides of the heart are separated by a wall called the **septum**. The right side of the heart collects blood from the body and pumps it to the lungs, whereas the left side receives blood from the lungs and pumps it to the rest of the body.

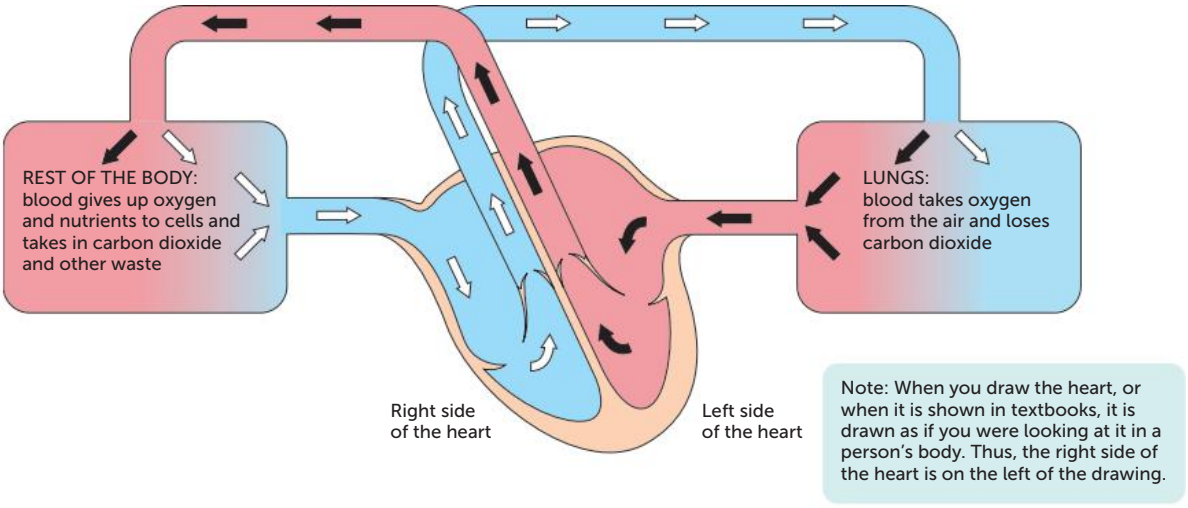


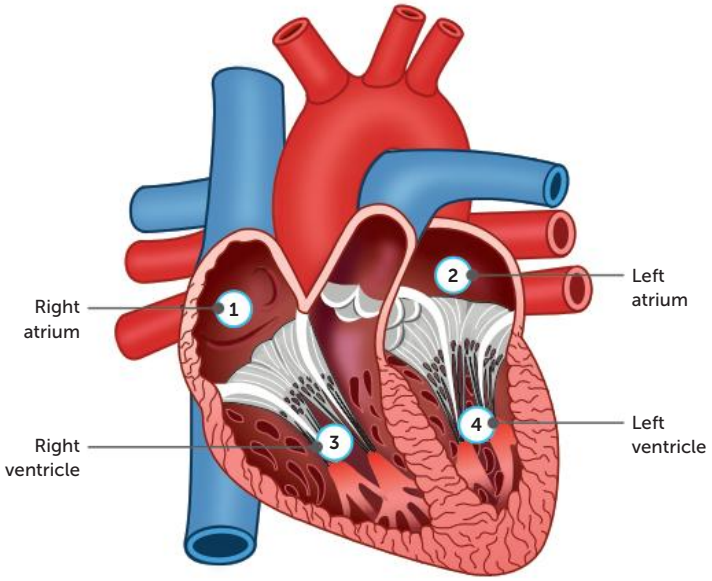
FIGURE 5.9 The flow of blood through the heart, body and lungs

Each side of the heart is also divided into two chambers; therefore, there are four chambers in the heart. The top chambers are called **atria** (singular: **atrium**), and the bottom chambers are the **ventricles**.

- The right atrium receives blood from the body and passes it to the right ventricle.
- The right ventricle pumps blood to the lungs.
- The left atrium receives blood from the lungs and passes it to the left ventricle.
- The left ventricle pumps blood to the body.

The wall of the left ventricle is thicker than the wall of the right ventricle. This is because it needs to be much stronger to pump the blood through the blood vessels supplying the body.

FIGURE 5.10
Simplified diagram of the chambers of the heart



9780170449090

Key concept

The heart is a four-chambered pump. The atria collect blood from the body and lungs, while the ventricles pump blood out of the heart to the lungs and body.

Valves in the heart

Valves in the heart ensure that the blood can only flow in one direction. Between the atria and the ventricles are the **atrioventricular valves**. These are flaps of thin tissue with the edges held by tendons, called **chordae tendineae**, that attach to the heart on **papillary muscles**. When the ventricles contract, the blood catches behind the flaps and they billow out like a parachute, sealing off the opening between the atria and the ventricles. Blood must then leave the heart through the arteries and not flow back into the atria.

Where the arteries leave the heart is a second set of valves that stop blood from flowing back into the ventricles when the ventricles relax. These are the **semilunar valves**. Each semilunar valve has three cusps. When blood flows into the artery, the cusps are pressed flat against the artery wall. When blood tries to flow back into the ventricle, the cusps fill out and seal off the artery, ensuring that the blood only flows in one direction. It is the closing of the valves that gives the heartbeats their characteristic 'lub dub' sound. The two sounds are due to the closing of the atrioventricular and then semilunar valves.

3D model of the heart
The beating heart

Key concept

Valves are located between the atria and the ventricles, and at the exit of the ventricles. They act to stop blood from flowing backwards.

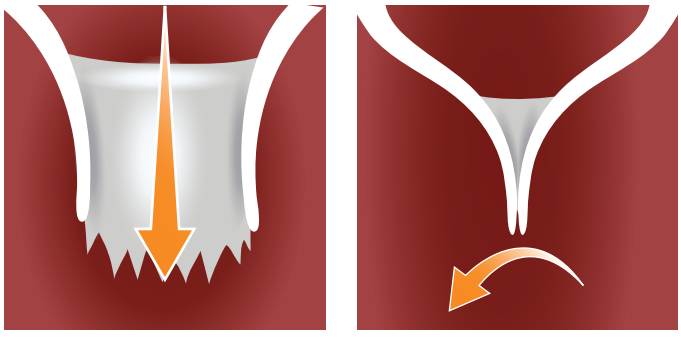
TABLE 5.2 Features of the valves of the heart

TYPE OF VALVE	NAME OF VALVE	LOCATION	NUMBER OF FLAPS OR CUSPS
Atrioventricular valve	Tricuspid valve	Between the right atrium and right ventricle	3
	Mitral (bicuspid) valve	Between the left atrium and left ventricle	2
Semilunar valve	Pulmonary valve	Between the right ventricle and pulmonary artery	3
	Aortic valve	Between the left ventricle and the aorta	3

Structure of the heart
Hover over the parts of the heart for more information about the structure.

Blood flow through the heart
Watch the video on this website to see the flow of blood through the heart.

Activity 5.3
Observing heart structure



Valve opens fully, blood flows through

Valve closes tightly, blood cannot flow backwards

FIGURE 5.11 Valves control the flow of blood by closing to prevent blood flowing backwards

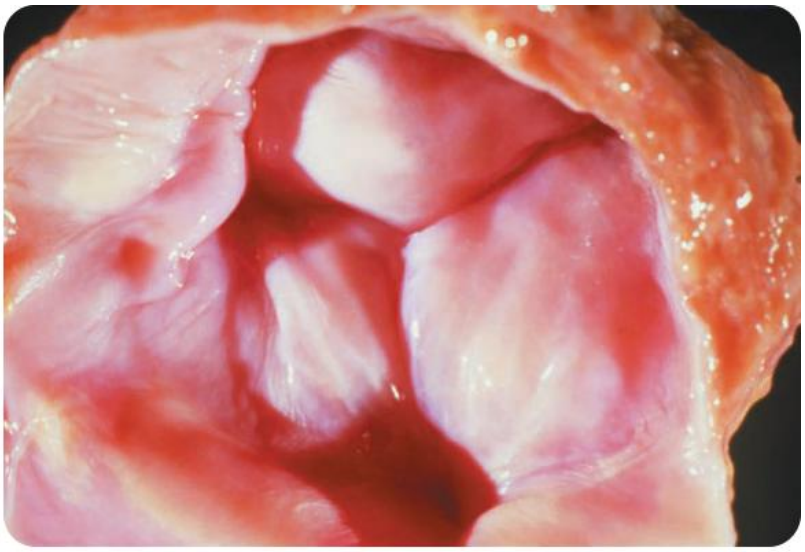


FIGURE 5.12
Tricuspid valve

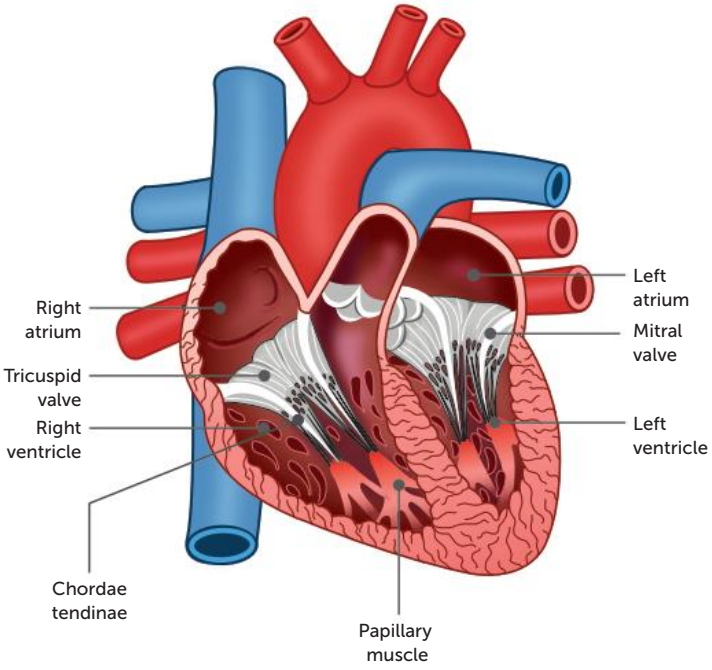


FIGURE 5.13 Valves are anchored by the chordae tendineae and papillary muscles

Blood vessels

Blood is pumped by the heart into blood vessels, which carry the blood to the cells of the body or the lungs, and then bring it back to the heart again. The same blood flows continuously through the heart, and this movement of blood is referred to as the **circulation**. There are three main types of blood vessels that are joined together to form the channels through which the blood flows: arteries, capillaries and veins.

9780170449090

Arteries

Arteries are the blood vessels that carry blood away from the heart. The largest artery is the aorta, which takes blood from the left ventricle to the body. Another important artery is the pulmonary artery, which takes blood from the right ventricle to the lungs.

The walls of an artery contain smooth muscle and elastic fibres. When the ventricles contract and push blood into the arteries, the walls of the arteries stretch to accommodate the extra blood. When the ventricles relax, the elastic artery walls recoil. This elastic recoil keeps the blood moving and maintains the pressure. The muscle in the artery walls *does not* contract and relax to pump the blood along. However, the muscle can contract to reduce the diameter of the artery and thus reduce blood flow to an organ. Such contraction of a blood vessel is called **vasoconstriction**. Conversely, the muscle may relax to increase blood flow to an organ in a process called **vasodilation**. In this way, blood flow may be controlled to allow for the changing needs of the body.

The very large arteries that receive blood pumped by the ventricles divide into smaller arteries. These in turn divide into very small arteries, known as **arterioles**. It is the arterioles that supply blood to the capillaries. Like the larger arteries, the arterioles have smooth muscle in their walls. Contraction or relaxation of this muscle is very important in regulating blood flow through the capillaries.

As the body exercises, the muscle cells continually require energy. Cellular respiration in the muscle cells makes the energy available, but also produces large amounts of wastes, including carbon dioxide and lactic acid. These wastes act as **vasodilators**, substances that produce a local widening, or dilation, of arterioles. This results in increased blood flow through the muscle tissues, ensuring that the cells are adequately supplied with oxygen and nutrients for continued functioning. Cellular respiration also releases heat energy, which tends to increase blood temperature. It also contributes to an increase in heart rate.

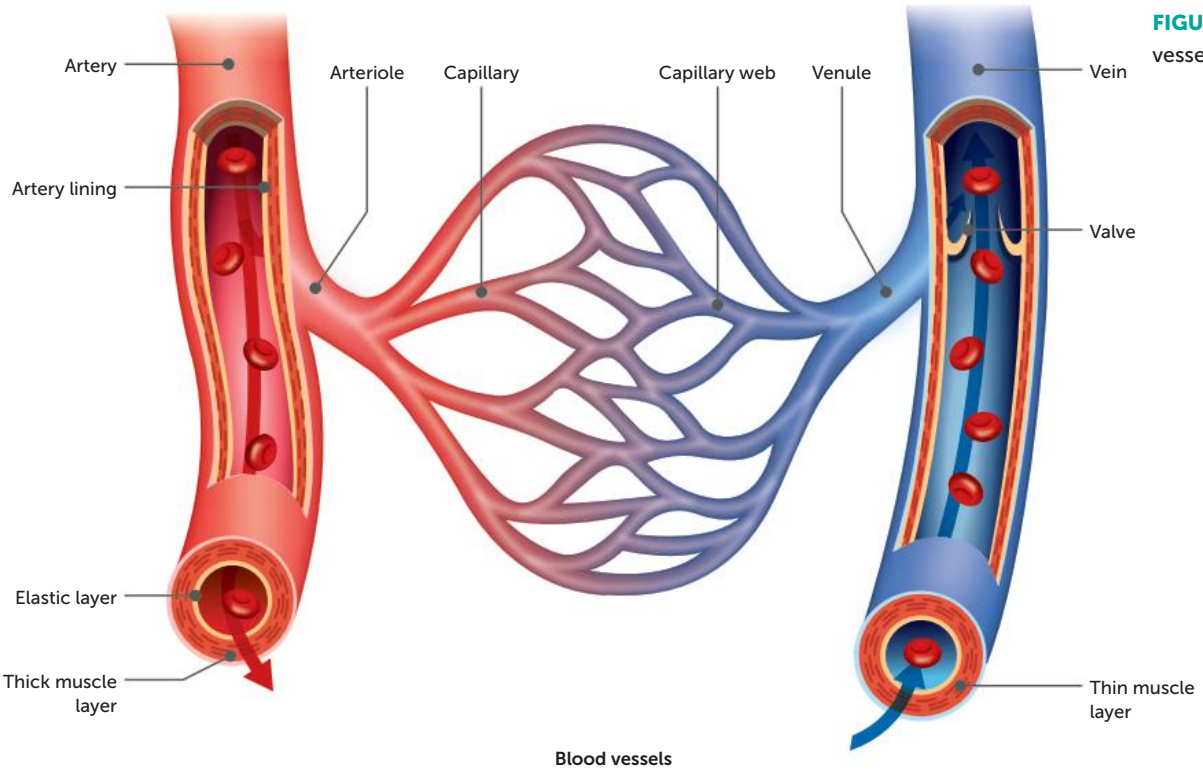


FIGURE 5.14 Blood vessels

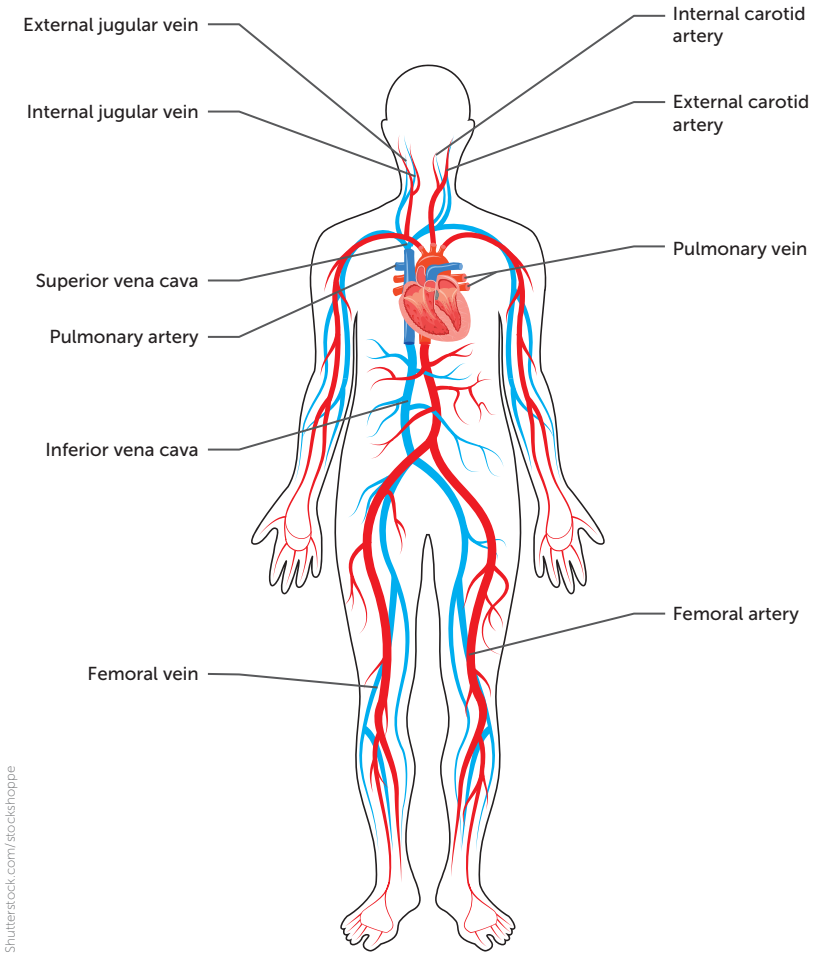


FIGURE 5.15 Major arteries and veins of the human body

Capillaries

Capillaries are the link between the arteries and veins. They are microscopic blood vessels that form a network to carry blood close to nearly every cell in the body. This enables the cells to get their requirements from the blood and to pass their waste into the blood. The structure of the capillaries makes them suitable for this function, as their walls have only one layer of cells. This allows substances to pass easily between the blood and the surrounding cells.

Activity 5.4
Observing capillaries

Veins

Veins carry blood towards the heart. The capillaries join into small veins, **venules**, which then join up to make larger veins. These culminate in the:

- **inferior vena cava** and **superior vena cava,** which bring blood from the body to the right atrium. The superior vena cava brings blood from above the heart, while the inferior vena cava brings blood from below the heart.
- **pulmonary veins**, which bring blood from the lungs to the left atrium. There are four pulmonary veins – two from each lung.

9780170449090

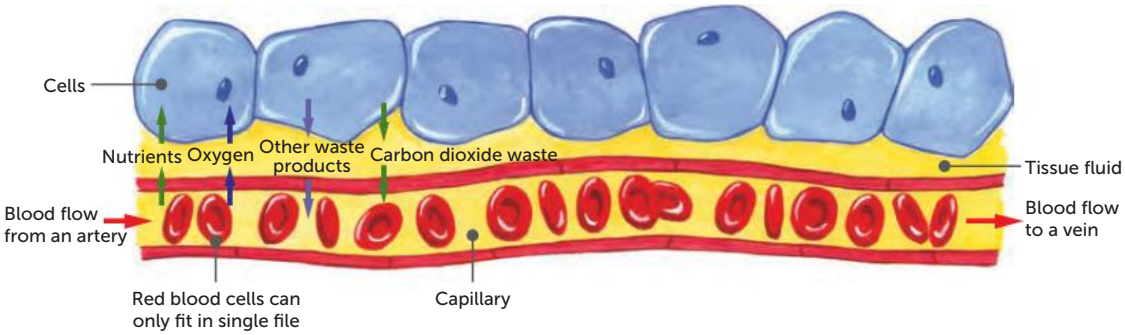
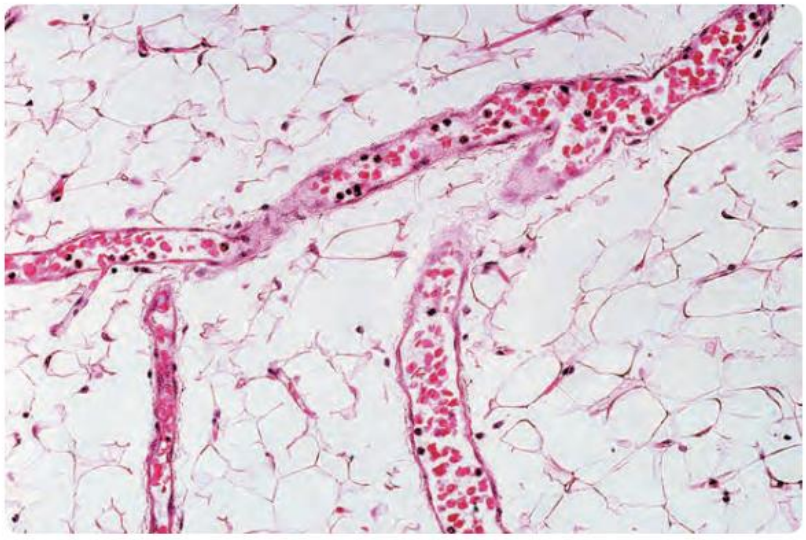


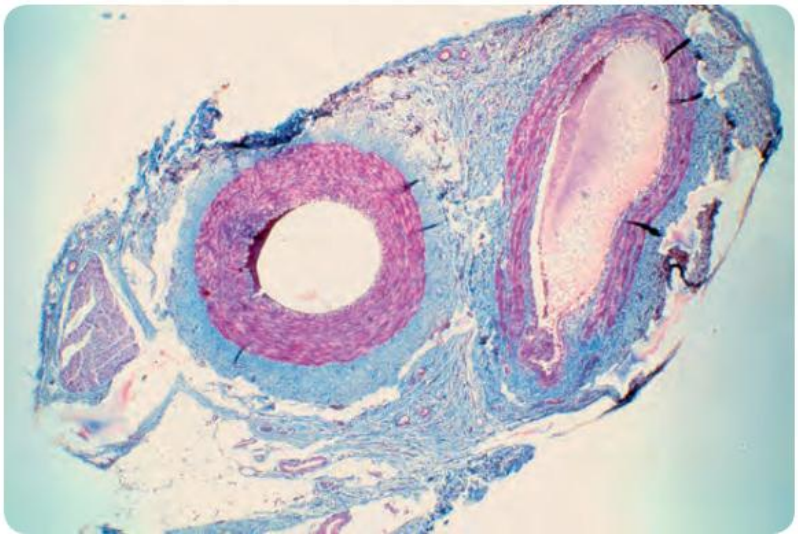
FIGURE 5.16 Capillaries allow the exchange of materials between cells and the blood



Alamy Stock Photo/Science Photo Library

FIGURE 5.17 Light micrograph of capillaries

Veins and venules do not have muscular walls and are not able to change their diameter in the way that arteries do. Blood pressure in the veins is relatively low because the blood loses most of its pressure as it flows through the tiny capillaries. The walls of veins are therefore much thinner than those of arteries; also, the pressure in veins is constant so the walls do not have to be elastic. Because of the low blood pressure, many veins have valves to prevent the blood from flowing backwards. Figure 5.19 shows how the valves in veins work.



Alamy Stock Photo/Science Photo Library

FIGURE 5.18 Transverse section of an artery (left) and a vein (right) showing the difference between the walls. The vein has a thinner wall and is being flattened by the surrounding tissue

9780170449090

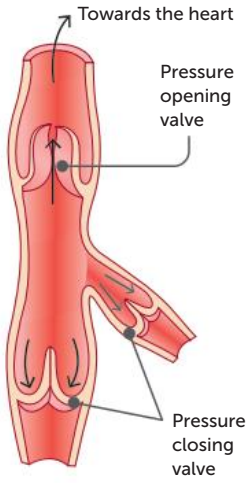


FIGURE 5.19 How the valves in a vein prevent backflow

Key concept

Arteries, capillaries and veins are the vessels through which blood travels around the body.

TABLE 5.3 Differences between arteries and veins

ARTERIES	VEINS
Carry blood away from the heart	Carry blood towards the heart
Have a blood pressure that increases as the ventricles contract and decreases as the ventricles relax	Have a constant, relatively low blood pressure
Have thick, muscular, elastic walls	Have thin, relatively inelastic walls with little muscle
Have no valves	Often have valves

WS

5.1 The circulatory system

Blood flow

As we have seen, blood is the transport medium that delivers oxygen and nutrients to cells and carries away their wastes. The requirements of cells vary depending on their level of activity. For example, when we are actively exercising, the muscles use up much more oxygen and nutrients, and produce more carbon dioxide and other wastes, than when we are sitting at rest. To cater for these changes in requirements, the blood flow to and from the cells must be able to change. There are two ways that this can occur:

- by changing the output of blood from the heart
- by changing the diameter of the blood vessels supplying the tissues.

Cardiac cycle

The **cardiac cycle**, or heartbeat, is the sequence of events that occurs in one complete beat of the heart. The pumping phase of the cycle, when the heart muscle contracts, is called **systole**. The filling phase, as the heart muscle relaxes, is called **diastole**. For a short time both atria and ventricles are in diastole. During this phase, the atria fill with blood and the ventricles also receive blood as the valves between them are open. **Atrial systole**, the contraction of the atria, then follows and forces the remaining blood into the ventricles. The atria then relax and refill while the ventricles contract in **ventricular systole**. Ventricular systole forces blood into the arteries. Although the left and right sides of the heart are two pumps, they operate together. Both atria contract simultaneously, as do both ventricles.

Activity 5.5
Investigating blood pressure

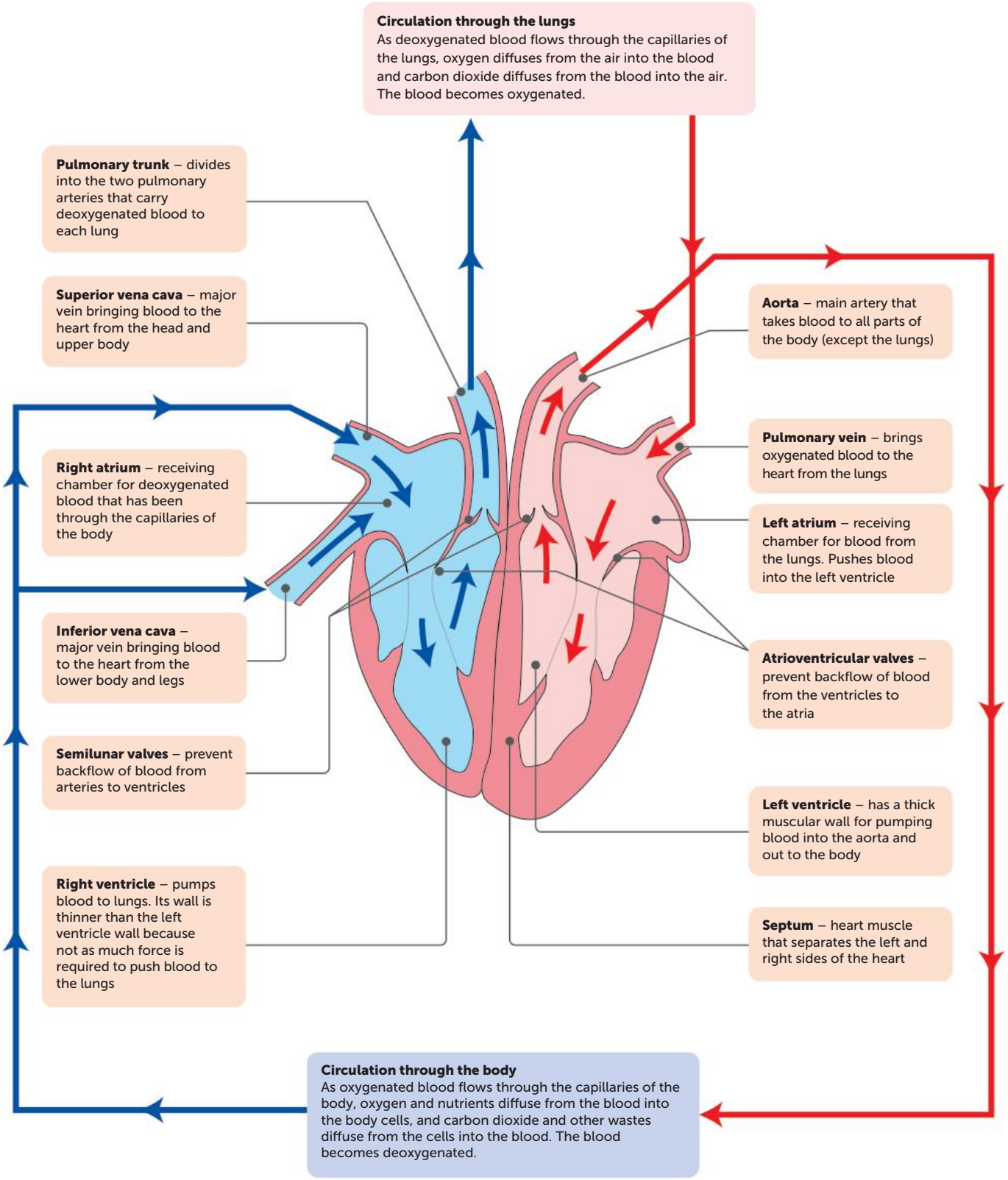


FIGURE 5.20 Structure of the heart and circulation of the blood

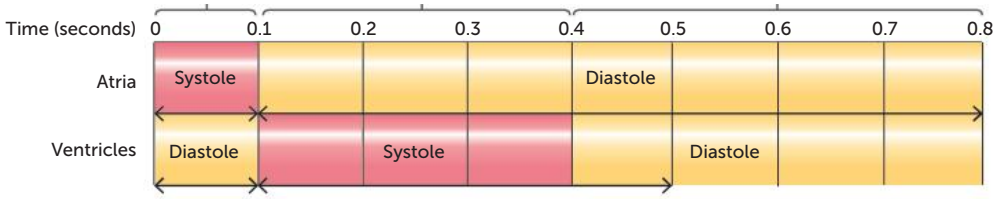


FIGURE 5.21 The cardiac cycle

Cardiac output

How quickly the blood flows around the body depends on how fast the heart is beating and how much blood the heart pumps with each beat. The **heart rate** is the number of times the heart beats per minute, while the **stroke volume** is the volume of blood forced from a ventricle of the heart with each contraction. A combination of both these factors influences the **cardiac output** – the amount of blood leaving one of the ventricles every minute. The cardiac output is equal to the stroke volume multiplied by the heart rate:

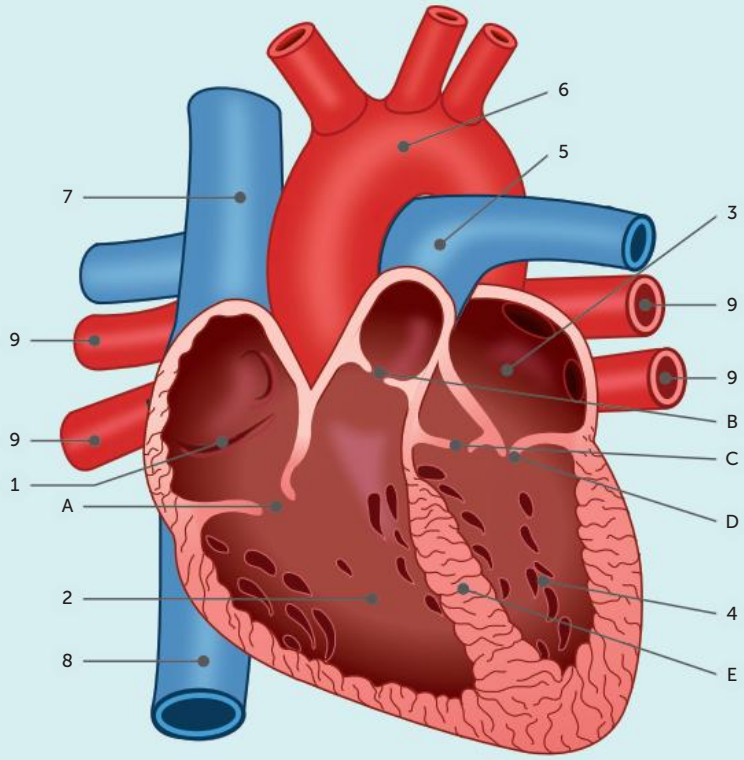
Cardiac output (mL/minute) = stroke volume (mL) × heart rate (beats/minute)

Circulation
This website contains more information on the blood, heart and blood vessels.

Questions 5.2

RECALL KNOWLEDGE

1 What type of blood vessel has the thickest walls?
2 Name the structure that stops the blood from travelling from the left ventricle to the left atrium.
3 Label the parts of the heart on the diagram below.



4 List the following structures in the order of blood flow through the body, starting from the lungs: Lungs, right atrium, left atrium, pulmonary vein, mitral valve, left ventricle, pulmonary valve, tricuspid valve, aorta, vena cava, pulmonary artery, aortic valve, right ventricle

5 Describe the function of capillaries.
6 Define 'cardiac output'.
7 Name the stage of the cardiac cycle when the atrium contracts.

APPLY KNOWLEDGE

8 Compare and contrast vasodilation and vasoconstriction.
9 Explain the importance of the papillary muscles.
10 Explain why the pulmonary artery carries deoxygenated blood, while other arteries carry oxygenated blood.
11 Mitral valve regurgitation is a condition resulting from a damaged mitral valve. Two of the symptoms of mitral valve regurgitation are an enlarged left atrium and shortness of breath during exercise. Explain why these symptoms occur.
12 CPR is taught during first-aid training to allow blood to continue to move through the body when the heart stops working. During this process, the lower sternum is pushed to a third of the depth of the chest. Explain how this manoeuvre is able to push blood through the body.
13 Discuss the importance of the septum in the heart.
14 Calculate the cardiac output if the heart rate is 58 beats per minute and 70 mL of blood is expelled with each beat.
15 Explain the difference between systole and diastole.

5.3 BLOOD GROUPS AND TRANSFUSIONS

A blood **transfusion** can be given to a person suffering from excessive blood loss, some types of anaemia, leukaemia, haemophilia or other conditions. It involves blood, or a blood product, from a donor being injected directly into the patient's bloodstream.

Some of the earliest, and unsuccessful, transfusions took place in the 17th century and involved the transfer of animal blood to humans. Early attempts at transfusions of whole blood between humans met with either spectacular success or the death of the patient.

In 1901, Karl Landsteiner (Figure 5.22), an Austrian doctor, experimented by mixing samples of blood from different people. His research led to the discovery of what is now called the **ABO blood group system**. Thirty-nine years later, Landsteiner, by then a citizen of the United States, discovered the **Rh blood group system.** A number of additional blood group systems have been discovered, by Landsteiner and others, but the ABO and Rh groupings are of particular importance in blood transfusions.

Blood groups

The surface of red blood cells is coated with sugar and protein molecules that are able to stimulate the immune system. These molecules are called **antigens** and the protein produced by the immune system is called an **antibody**. The antigen and its antibody combine to form a complex and cause a reaction.

Activity 5.6
Investigating blood typing

ABO blood groups

There are two sugar antigens involved in the ABO classification of blood groups: antigen A and antigen B. On the surface of the red blood cells a person may have either antigen A, antigen B, both antigens or neither antigen. These four possibilities correspond to the four groups of the ABO system: group A (antigen A), group B (antigen B), group AB (both antigens) and group O (neither antigen). The body's ability to make the antigens, and so a person's ABO blood group, is determined by their DNA and is therefore inherited.

9780170449090

FIGURE 5.22
Karl Landsteiner discovered the ABO and Rh human blood group systems

WS

5.2 Blood groups

The antibody that reacts against antigen A is called anti-A, while anti-B reacts against antigen B. A person's immune system is able to recognise their own antigens and will not produce antibodies for them. However, they will produce antibodies for antigens that are non-self. Thus, a group A person can produce only the antibody anti-B, a group B person can produce only anti-A, a group AB person cannot produce either antibody, and a group O person can produce both. This is summarised in Table 5.4 on page 122.

Rh blood groups

The Rhesus blood group system is so named because Landsteiner used the blood of rhesus monkeys in his initial investigations. Like the ABO system, it is based on antigens that occur on the surface of the red blood cells. Unlike the ABO antigens, which are sugars, the Rh antigens are proteins.

A person with Rh antigens is said to be Rh positive; a person without these antigens is Rh negative. An individual without the Rh antigens is able to produce an anti-Rh antibody that reacts against those antigens. Rh-positive individuals cannot produce an anti-Rh antibody.

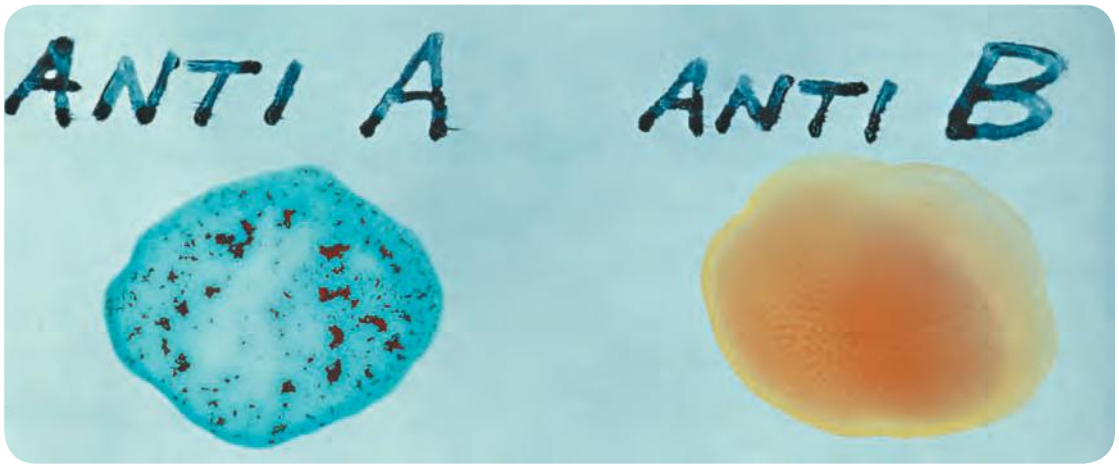
Key concept

Red blood cells contain antigens on their surface. These antigens determine the blood group of the individual, including their ABO and Rh blood groups.

Transfusions

As indicated previously, a transfusion transfers blood, or one of the components of blood, from one person to another. For most transfusions it is necessary to match the blood groups of the donor and the recipient, although the use of some blood products, such as clotting factors, may not require matching of blood groups.

The mixing of blood types that are incompatible can cause the erythrocytes to clump together, or **agglutinate**. If the receiver's blood contains, or is able to make, antibodies against the antigens on the donor's red cells, the foreign cells will clump together and disintegrate. It is therefore essential that the blood group of the receiver and donor be the same. The ABO blood group of the donor is always matched to that of the receiver when transfusions are given (see Table 5.4).



Getty Images/Ed Reschke

FIGURE 5.23 Mixing blood. When mixed with incompatible plasma (type B) (left), the red cells clump together. When mixed with compatible plasma (type B) (right), no clumping of red cells occurs

Rh blood groups are also matched for transfusion purposes. The anti-Rh antibody is not normally present in the plasma of Rh-negative people, but it is produced on exposure to the Rh antigen. The first transfusion of Rh-positive blood to an Rh-negative patient does not usually cause problems because the antibodies are produced slowly. However, that first exposure sensitises the person, so that any subsequent exposure results in very rapid production of antibodies. Clumping of the red cells results, in a manner similar to ABO incompatibility.

Types of transfusions

Whole blood is blood as it is taken from the donor but with a chemical added to prevent clotting. Transfusions of whole blood are used mainly in cases of severe blood loss.

Red cell concentrates are the most widely used component of blood. They are produced by spinning blood at very high speed in a centrifuge. The heavier cells sink to the bottom, leaving the lighter plasma on top. The concentrate may or may not have platelets and white blood cells (leucocytes) removed. Transfusions of red cell concentrates are used for patients suffering from heart disease or severe anaemia.

Plasma, the liquid part of the blood, may be given to patients requiring extra clotting factors for control of severe bleeding, or to patients with liver disease.

Platelet concentrates are given to patients who have abnormal platelets or a reduced number of platelets.

Cryoprecipitate is obtained by freezing the plasma and thawing it slowly. When the plasma is thawed, the cryoprecipitate remains solid. It contains many of the substances necessary for blood clotting. Cryoprecipitate may be used to treat some forms of **haemophilia**, but it is most often used for severe bleeding.

Immunoglobulins are a group of proteins that act as antibodies. They are extracted from the blood and used for patients who are deficient in antibodies. Particular immunoglobulins from certain donors are used to treat patients who have no immunity to a particular disease. For example, tetanus immunoglobulin may be used to treat tetanus.

An **autologous transfusion** is when the patient's own blood is used. The blood is collected from the patient prior to an operation that may require a transfusion. Such transfusions are often used for elective surgery and the blood is collected about four weeks before the operation. Autologous transfusions eliminate the risk of transmission of disease and most possible side effects of the usual transfusions.

Key concept

Different types of blood transfusions are used for different conditions.

9780170449090

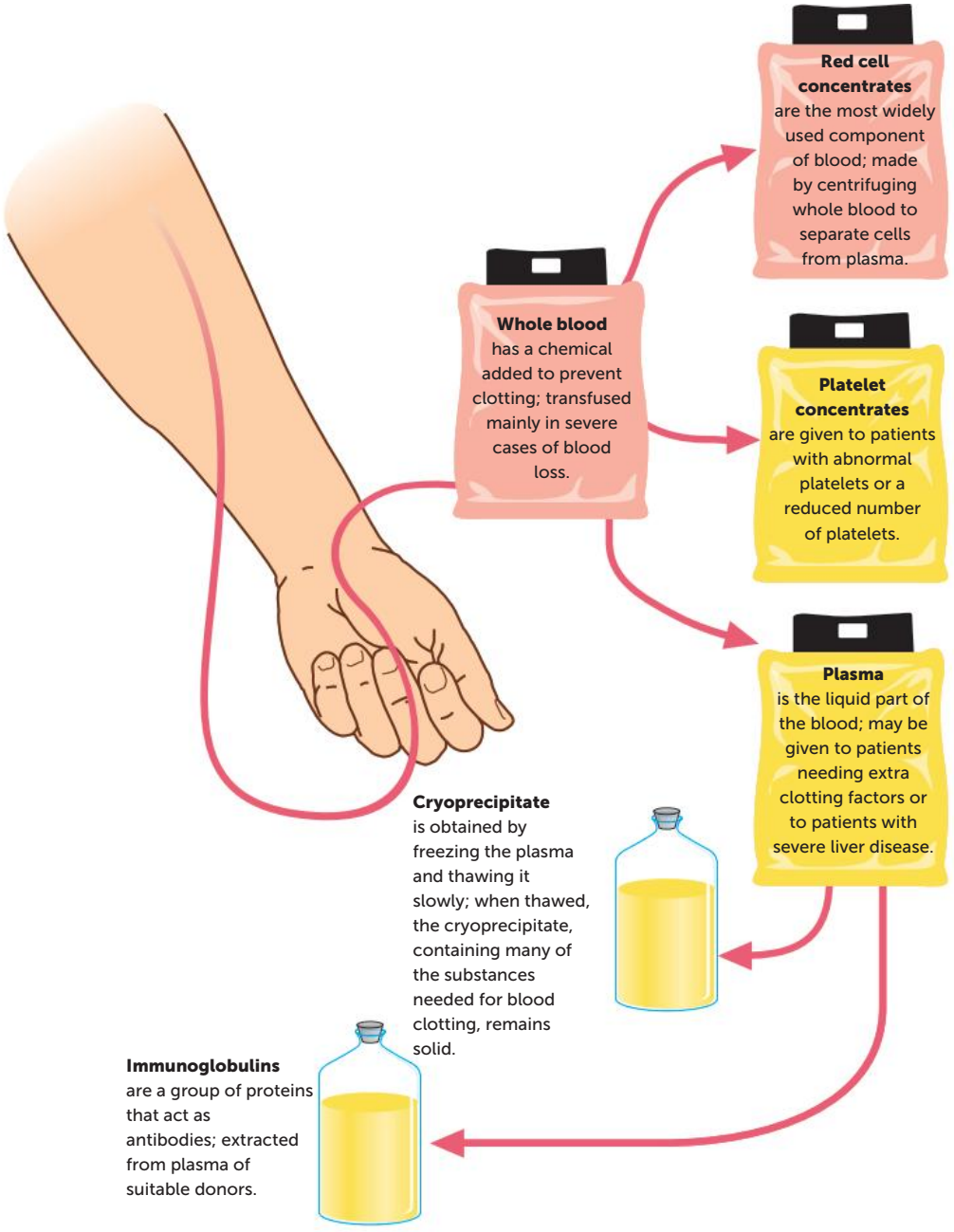


FIGURE 5.24 Blood and blood products used for transfusion

TABLE 5.4 Summary of ABO blood groups and donor–recipient matching

BLOOD GROUP	ANTIGENS ON RED BLOOD CELLS	ANTIBODIES IN PLASMA	ABLE TO DONATE TO PEOPLE WITH BLOOD GROUP ...	ABLE TO RECEIVE BLOOD (RED CELL CONCENTRATE) FROM PEOPLE WITH BLOOD GROUP ...
A	Antigen A	Anti-B	A, AB	A, O
B	Antigen B	Anti-A	B, AB	B, O
AB	Antigen A and antigen B	Neither anti-A nor anti-B	AB	A, B, AB, O
O	Neither antigen A nor antigen B	Both anti-A and anti-B	A, B, AB, O	O

Blood transfusion donor and recipients
Click on the donor blood type and see which recipients can receive it.

Blood typing
Find out more about blood groups at this website.

9780170449090

Becoming a blood donor

In Australia, blood is free. It is collected from donors by the Red Cross Blood Transfusion Service and no charge is made for the blood or for blood products. Most states and territories in Australia have laws that prohibit payment for blood donations or charges for blood supplied to patients.

The Red Cross always needs blood donors and at times there are critical shortages of certain types of blood. Any healthy person aged 16–70 years can be a potential donor, although in some states 16- and 17-year-olds may require parental permission.

Donating blood is a worthwhile way of contributing to society. It costs nothing and gives the donor the satisfaction of helping others.

Alamy Stock Photo/BSIP SA

FIGURE 5.25
Donating blood

Questions 5.3

RECALL KNOWLEDGE

1. List four reasons that someone would require a blood transfusion.
2. List the antibodies that would be present or produced from someone with B-positive blood.
3. Describe the process of agglutination.
4. What is the most commonly transfused part of blood?

APPLY KNOWLEDGE

5. Explain the importance of matching blood groups between the donor and recipients of a blood transfusion.
6. Blood group O is known as the universal donor. Explain why this is valid.
7. Compare and contrast the ABO blood groups and the Rh blood groups.
8. Explain why patients with severe bleeding receive plasma transfusions.

5.4 THE LYMPHATIC SYSTEM

As blood enters the capillaries, the relatively high pressure forces some of the fluid in the blood through the capillary walls into the tissues. The main function of the **lymphatic system** is to collect some of the fluid that escapes from the blood capillaries and return it to the circulatory system. The lymphatic system is also an important part of the body's internal defence against disease-causing organisms.

Structure of the lymphatic system

The lymphatic system consists of:

- a network of lymph capillaries joined to larger **lymph vessels** (also called **lymphatic vessels** or **lymphatics**)
- lymph nodes, which are located along the length of some lymph vessels.

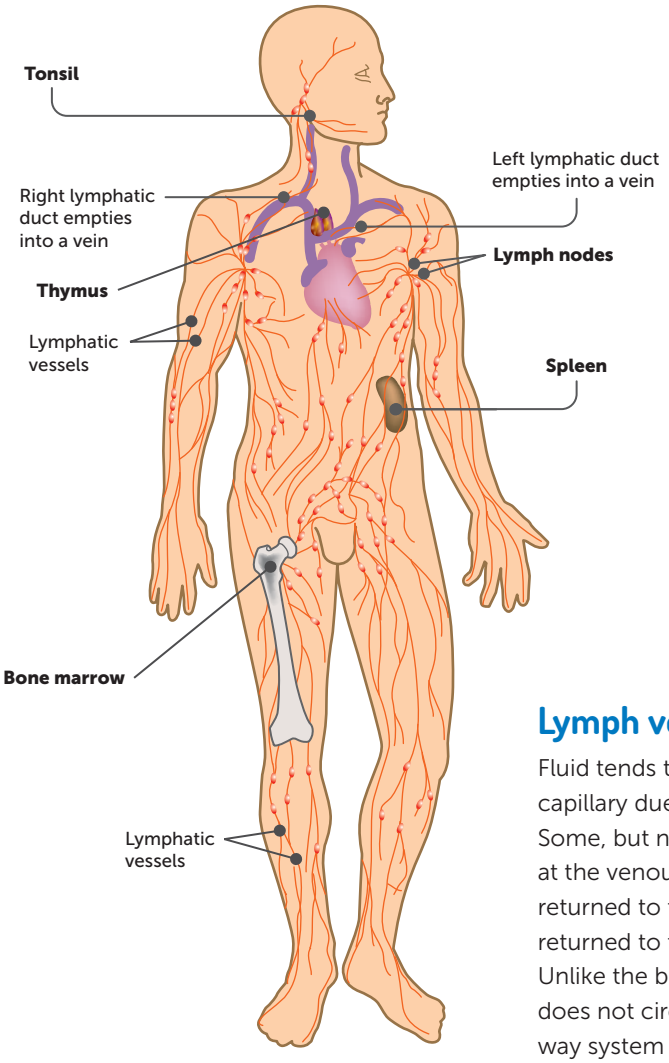


FIGURE 5.26 Structure and function of the lymphatic system

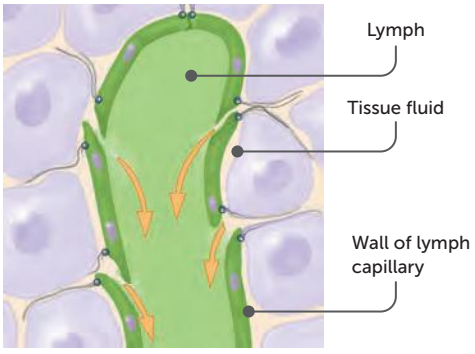


FIGURE 5.27 Lymph capillaries have blind ends

Lymph vessels

Fluid tends to leak out at the arterial end of a blood capillary due to the high pressure in the vessel. Some, but not all, of this fluid returns to the capillary at the venous end. The excess fluid in the tissues is returned to the blood by the lymphatic system. Fluid returned to the blood in this way is known as **lymph**. Unlike the blood in the circulatory system, lymph does not circulate – the lymphatic system is a one-way system carrying fluid away from the tissues. The lymph vessels originate as blind-ended tubes in the spaces between the cells of most tissues.

Lymph capillaries are usually slightly larger than blood capillaries. They are also more permeable than most blood capillaries. Proteins and disease-causing organisms in the **intercellular fluid** can easily pass through the walls of the lymph capillaries into the lymph.

The network of lymph vessels joins to form two lymphatic ducts that empty the lymph into large veins in the upper chest.

9780170449090

Lymph is moved through the lymphatic vessels as a result of smooth muscle, skeletal muscle and valves. The smooth muscle layer of the vessels is able to contract to push the lymph along the vessel. The skeletal muscles surrounding the vessels are also able to contract, providing additional force. As there is no central pump, there is no force driving the direction of the flow of lymph. Therefore, the larger lymph vessels have valves that close when the pressure drops, preventing the backflow of lymph.

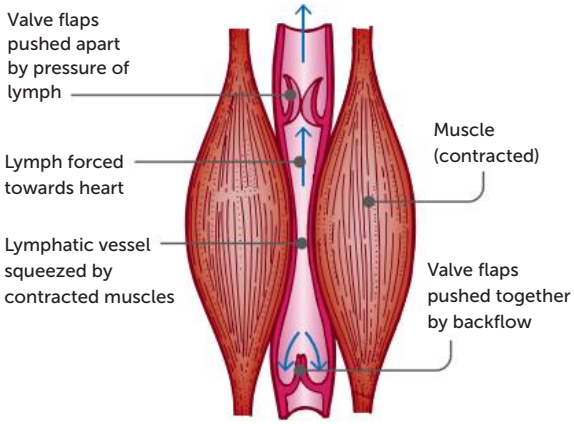


FIGURE 5.28 Lymph is pushed along the lymph vessels by the squeezing action of skeletal muscles. Valves prevent the backflow of lymph

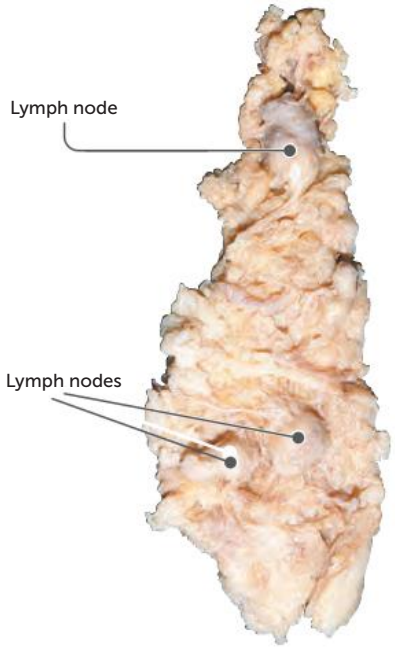
Key concept

Lymph is fluid that is collected from between the cells and transported in lymph vessels to the large veins.

Lymph nodes

Lymph nodes, also called **lymph glands**, occur at intervals along the lymphatic vessels. They are most numerous in the neck, armpits, groin and around the alimentary canal. Nodes are bean-shaped and range in length from 1 mm to 25 mm. Each is surrounded by a capsule of connective tissue that extends into the node, forming a framework. Within the framework are masses of **lymphoid tissue**, containing cells known as **lymphocytes**, **macrophages** and **plasma cells**. Spaces between the cells of the lymphoid tissue are criss-crossed by a network of fibres.

Lymph enters through vessels on the convex side of the node, filters through the spaces and passes out through vessels on the opposite side. The lymph passes through several nodes before entering the circulatory system.



From Saladin K. ed. *Anatomy and Physiology: The Unity of Form and Function*. 3rd edn. McGraw-Hill. 2004.

FIGURE 5.29 Lymph nodes embedded in connective tissue

Role of the lymphatic system in defence against disease

Lymph entering the lymph nodes contains cell debris, foreign particles and micro-organisms that have penetrated the body's external defences. Some of these micro-organisms may be able to cause disease and must therefore be destroyed.

Larger particles, such as bacteria, are trapped in the meshwork of fibres as the lymph flows through the spaces in the nodes. Large **phagocytic cells** called macrophages destroy these particles. The macrophages ingest the particles by phagocytosis. Projections from the macrophage surround the particle and take it into the cell, where it is destroyed by enzymes. Most bacteria ingested in this way are killed within 10 to 30 minutes.

When infections occur, the formation of lymphocytes increases, causing the lymph nodes to become swollen and sore. For example, an infected finger may result in swelling and tenderness in the armpit, where there are a large number of lymph nodes.

Key concept

Lymph nodes contain cells that help protect the body, such as macrophages, which engulf foreign particles, and lymphocytes, which are part of an immune response.

Lymphatic system

9780170449090

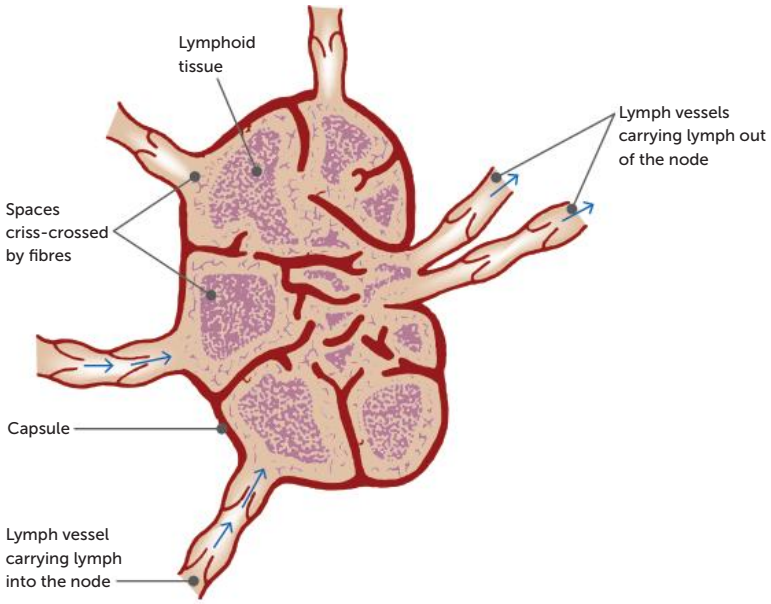
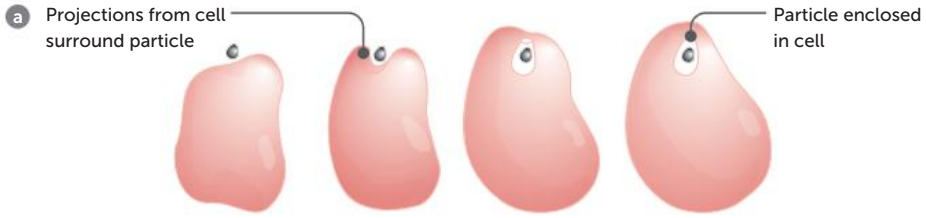


FIGURE 5.30 Section through a lymph node; the arrows indicate the direction of lymph flow



Science Photo Library/Steve Gschmeissner

FIGURE 5.31
a The process of phagocytosis;
b Macrophages attacking a large foreign particle

Questions 5.4

RECALL KNOWLEDGE

1. Define 'lymph'.
2. Use a flow chart to show the movement of lymph from the blood capillaries to the veins in the upper chest.
3. List the functions of the lymphatic system.
4. Describe the role of lymph nodes in protecting the body against foreign bodies.

APPLY KNOWLEDGE

5. Discuss the importance of valves in lymph vessels.
6. Doctors will check the size of lymph nodes in sick patients. Explain why this is an important part of the physical examination.
7. Compare and contrast lymph vessels and blood vessels.

9780170449090

CHAPTER 5 ACTIVITIES

ACTIVITY 5.1 Comparing blood cells

Examining blood with a microscope will help you to appreciate and remember the structure of blood.

You will need

Microscope; microscope lamp; prepared slide of blood smear; minigrid

What to do

1. Examine a prepared slide of a blood smear. The blood cells on this slide will have been stained. Observe the cells first on low or medium power, then gently change to high power. Identify each of the types of cells that you can see.
2. Use a minigrid to determine the field diameter of your microscope on low or medium power (see Activity 2.1 and the figure on page 50 in Chapter 2). Calculate the field diameter on high power and then estimate the diameter of a red blood cell.

Studying your observations

1. Describe the appearance of a red blood cell. What does the appearance tell you about the structure of the cell?
2. Draw a diagram showing a red blood cell and each of the types of white blood cells that you can see.
3. What is the approximate difference in size between red blood cells and white blood cells?
4. What is the approximate ratio of numbers of red blood cells to white cells?
5. Were you able to see any platelets? Suggest why platelets are difficult to see with a school microscope.

ACTIVITY 5.2 Investigating blood flow during exercise

The table below shows blood flow through various parts of the body when a person is sitting at rest and during moderate exercise.

PART OF THE BODY	RATE OF BLOOD FLOW (mL/min)	
	RESTING	EXERCISING
Skeletal muscle	1 000	12 500
Digestive system (stomach, intestines, liver)	1 350	600
Kidneys	1 100	600
Brain	700	750
Skin	300	1 900
Heart muscle	200	750
Other organs	350	400

Source: Data from Saladin K, ed. *Anatomy and Physiology: The Unity of Form and Function*. 3rd edn. New York: McGraw-Hill, 2004.

Your task

1. Calculate the percentage increase in blood flow through each organ during exercise.
2. Construct a graph to show the percentage increase in blood flow for each organ.

Discussion

Use the data in the table, or your graph, to answer the following questions.

1. Calculate the person's cardiac output when at rest.
2. Calculate the person's cardiac output while exercising.
3. What are the two ways in which cardiac output can be increased?
4. For each part of the body listed in the table, explain the reasons for any changes in blood flow that occur during exercise.
5. Which body organ experiences the greatest increase in blood flow during exercise? Explain the reason for this.

ACTIVITY 5.3 Observing heart structure

Although the hearts of mammals differ greatly in size, they are all basically similar. This activity will help you to learn about the structure and functioning of your own heart.

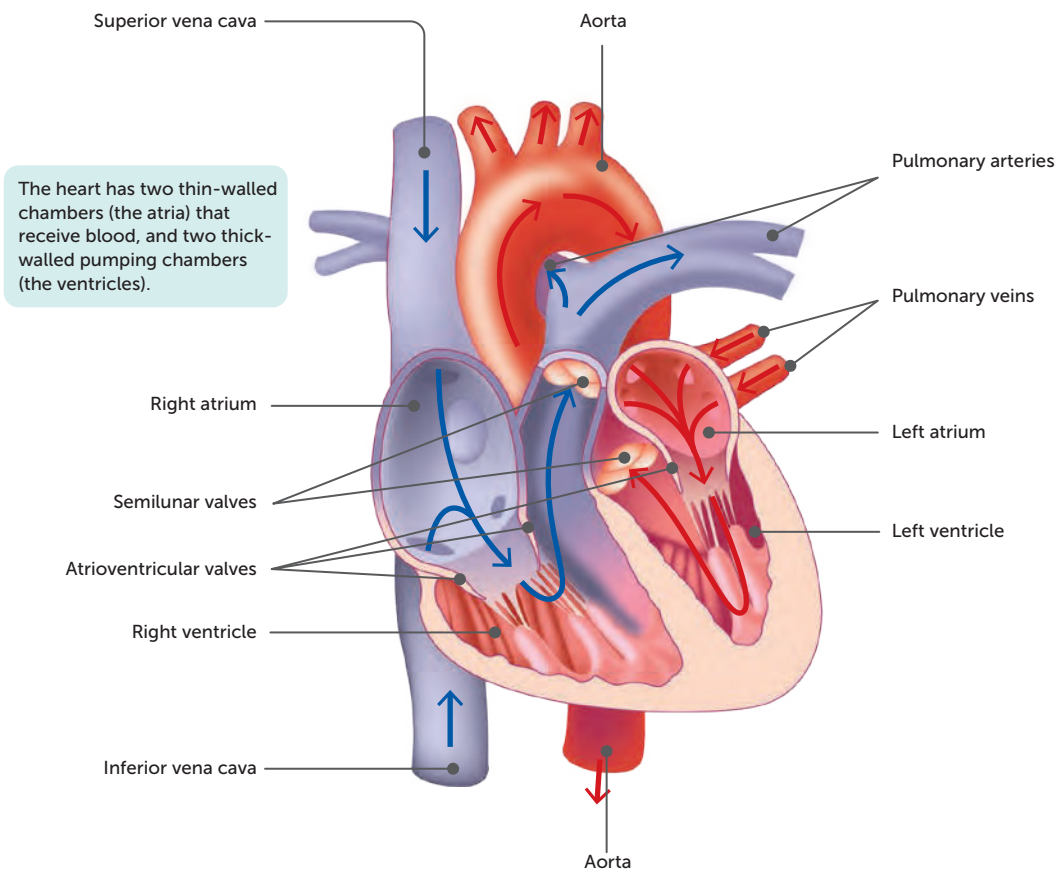
Your teacher may wish you to dissect a mammal heart yourself, may demonstrate the dissection, or may refer you to a video for this activity.

You will need

Heart of a sheep, pig or cow; dissecting board; dissecting instruments; disposable gloves

What to do

Refer to the figure below as you do the activity.



1 Identify the four chambers of the heart. Part of each atrium may have been removed when the heart was cut from the animal.
2 Identify the left and right sides of the heart. Feel the ventricle walls – the left ventricle has a much thicker wall and feels much firmer than the right ventricle. Also, the tip of the heart (the apex) is part of the left ventricle.
3 Identify the aorta and the artery to the lungs. Look for the pulmonary veins and venae cavae (veins from the body), although these might have been removed by the butcher.
4 Complete step 1 of 'Studying your observations' before continuing.
5 Follow your teacher's instructions to cut open the heart. This involves identifying the groove on the outside of the heart that marks the division between the left and right ventricles, then cutting open the left ventricle along a line parallel to and about 2 cm from the groove.
6 Open the ventricle and locate the flaps of the valve between the atrium and the ventricle. Note the tendons attached to the edges of the flaps.
7 Continue your cut through the wall of the atrium and through the wall of the aorta. Open out the aorta and locate the three cusps of the semilunar valve. These will be close to where the aorta leaves the ventricle. You may have cut through one of the cusps.
8 Open the right side of the heart in a similar way and identify all the structures on that side.

Studying your observations

1 Arrange the heart as you would see it if you were looking at it in a person's chest. Take a photo or draw the heart and label all the external features.
2 Measure the thickness of the wall of each of the four heart chambers. List the chambers in order from that with the thinnest wall to that with the thickest wall.
3 In your own words, describe the appearance of the atrioventricular valves. Are there any differences between the left and right atrioventricular valves?
4 Describe the appearance of the semilunar valves. Are there any differences between the semilunar valves of the aorta and those of the pulmonary artery?
5 Describe any differences that you observed between the veins and the arteries.
6 Why does the heart have two types of chamber – atria and ventricles?
7 Why does the heart have two of each chamber – two atria and two ventricles?

ACTIVITY 5.4 Observing capillaries

Your teacher may demonstrate, or you may observe for yourself, the blood flowing through capillaries in the tail of a small fish such as a goldfish or gambusia.

1 Place the fish in half a Petri dish and cover the front half of the animal with wet cotton wool. Put the Petri dish on the stage of a microscope. Using the lowest magnification, focus on the tip of the fish's tail. Do not keep the fish out of water for more than three minutes.
2 Describe what you observe.
3 How does the diameter of a blood cell compare with the diameter of the smallest capillary?

9780170449090

ACTIVITY 5.5 Investigating blood pressure

The heart is responsible for pumping blood throughout the cardiovascular system. The pressure of the blood that the heart pumps through the arteries and veins is not consistent. With each heartbeat, the blood pressure surges. When the heart beats, the blood pressure in the vessels is the highest, and is at its lowest when the heart is relaxed. 'Blood pressure' describes the force created by the blood against the inner walls of the blood vessels.

To monitor blood pressure, medical professionals commonly use a sphygmomanometer, an instrument that is calibrated in pressure units that indicate systolic pressure and diastolic pressure. As the ventricles contract to squeeze the blood inside their chambers, the pressure in the arteries rises sharply. This pressure, called systolic pressure, is the maximum pressure achieved during ventricular contraction. When using a sphygmomanometer, this pressure can be heard as faint rhythmic thumping sounds as the blood returns to the arm when the sphygmomanometer cuff is slowly deflated. As the ventricles relax, the arterial pressure drops. This pressure, called diastolic pressure, is the lowest pressure that remains in the arteries before the next ventricular contraction.

Aim

To measure blood pressure while resting and to test the effects of exercise on blood pressure.
Time requirement: 30 minutes

You will need

Sphygmomanometer; stethoscope; clock or timer

Risks

WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
Be aware of physical considerations such as heart- or health-related problems	Ensure that students with any known physical conditions do not participate in the physical part of the investigation.

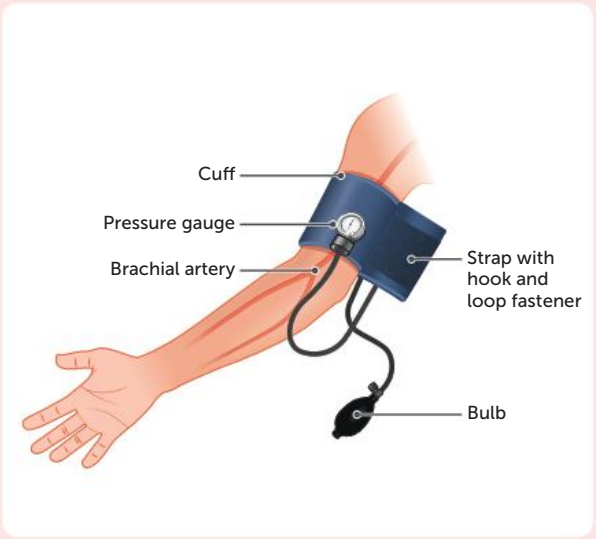
What to do

Preparing the pressure cuff

1. Working in pairs and taking turns, become familiar with the parts of the sphygmomanometer and stethoscope. Place the stethoscope around your neck, as you will need it shortly.
2. Inspect the sphygmomanometer – a device used to measure blood pressure. You should observe that a cloth-covered rubber cuff forms the main body of the sphygmomanometer. Attached to this cuff via rubber tubes are two objects: the hand bulb and the pressure gauge.
3. Ask your partner to roll up their right sleeve until it reaches past their elbow.
4. Also ask them to extend their right arm with the palm facing upwards. For accurate readings, the arm needs to be at the same level as the heart. This can be achieved by the person lying down or sitting with their arm resting on a table.
5. Find the large brachial artery near the hollow of the elbow by feeling for the pulse of the artery. You will use this artery to monitor the heartbeat.
6. Carefully place the deflated pressure cuff on your partner by wrapping it around the upper arm so that the lower edge of the cuff is approximately 2.5 cm above the elbow.

9780170449090

7 Ensure the cuff is wrapped closely around the arm without being too tight. Once you are satisfied with the fit, secure the cuff using the hook and loop tape.



Taking blood pressure

1 Using your fingertips, feel the pulse of the artery and close the valve located on the bulb by rotating it clockwise.
2 Inflate the cuff by squeezing the bulb. Continue to inflate until you can no longer feel the pulse. Stop inflating immediately if your partner becomes uncomfortable or faint or experiences any pain.
3 To deflate the cuff, turn the valve on the bulb counterclockwise. As you deflate the cuff, keep your fingertips on the artery and monitor the gauge.
4 Record the point on the gauge at which you can feel the pulse in the artery once again.
5 Wait for the cuff to deflate completely. After 30 seconds' rest, repeat this procedure.
6 This time, inflate the cuff until the gauge reaches the point you previously noted. After you have reached this point, continue to inflate the cuff an additional 30 mm.
7 As you slowly deflate the cuff, secure the earpieces of the stethoscope in your ears and place the flat side where the brachial artery is located. Gradually deflate the cuff by turning the valve on the bulb counterclockwise.
8 Practise deflating slowly until you are able to drop the pressure 2–4 mm with each heartbeat. This would be approximately 1 to 2 marks on the gauge each second. It is important to maintain this rate of deflation to gain accurate readings. Be aware that the cuff cuts off blood flow and should not be left inflated any longer than is absolutely necessary.
9 As you slowly open the valve, listen for the faint rhythmic thumping sounds as the blood returns to the arm. Note the reading on the gauge immediately when you hear these faint noises. This figure is the systolic (upper) blood pressure reading.
10 Once again, release the pressure slowly from the cuff so that it continues to drop at 2–4 mm per second. Using the stethoscope, listen for sharp tapping sounds that soften to blowing or swishing sounds. As you listen, observe the falling gauge needle. Note the figure on the gauge at the precise point you are no longer able to hear any sounds. This number reveals the diastolic (lower) blood pressure reading.
11 Record your readings using the correct format. Note that blood pressure is written as a fraction, with systolic pressure over diastolic pressure – for example, 115/70 or 120/80.

9780170449090

Blood pressure and exercise

Before conducting this procedure, consider if you have any heart or other health problems. Inform your teacher immediately if you do. You may wish to consult a physician before taking part in this procedure. Stop at any point if you feel faint.

1. Complete this procedure working with your partner and taking turns. Measure your partner's blood pressure at rest after 5 minutes in a sitting position. Record your results in the table below (before exercise).
2. Ask your partner to run on the spot for 2 minutes, taking approximately two steps per second.
3. Measure their blood pressure immediately following the run, 2 minutes after exercise, and continue to take measurements every following 2 minutes until the blood pressure measurements return to the original figure that was taken at rest.
4. Record the blood pressure readings in the table below.

Heart rate and breathing rate

1. Form a group of four to allow you to take measurements of blood pressure, heart rate and respiration at the same time.
2. Repeat the 'Blood pressure and exercise' procedure, but this time assign somebody to monitor heart rate, and assign another person to monitor respiration.
3. Once the person has finished running, their pulse can be measured by locating the heartbeat at the wrist and counting the heartbeats for 15 seconds. Multiply the number of heartbeats in 15 seconds by 4 to generate the beats per minute.
4. Respiration can be measured by listening and recording the number of breaths taken in a minute.
5. Record the data in the table below.

Studying your results

Copy and complete the following table.

TIME	BLOOD PRESSURE (SYSTOLIC/DIASTOLIC)	PULSE (BEATS PER MINUTE)	RESPIRATION (BREATHS PER MINUTE)
Before exercise			
Immediately after exercise			
2 minutes after exercise			
4 minutes after exercise			
6 minutes after exercise			
8 minutes after exercise			

9780170449090

Discussion

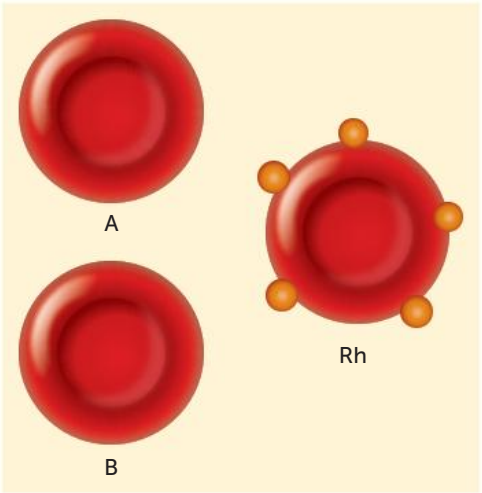
1 Describe how blood pressure is measured by the technique you used and the sphygmomanometer.
2 In this procedure, you were asked to listen to the systolic pressure. Describe what you were able to hear and how this corresponds with what systolic pressure is.
3 You were also asked to listen to the sounds of the diastolic pressure. Describe what you were able to hear and how this corresponds with what diastolic pressure is.
4 Describe what blood pressure is and how it occurs.
5 What are the potential consequences of prolonged high blood pressure?
6 How can hypertension (high blood pressure) be prevented?

SOUTHERN Biological Developed by Southern Biological

ACTIVITY 5.6 Investigating blood typing

Aim

To determine an unknown blood type in a sample of synthetic blood using the presence or absence of agglutination as evidence.



Example of an O+ blood type. Note that there are no A or B antigens.

Time requirement: 30 minutes

You will need

1 vial of sample 1 synthetic blood; 1 vial of sample 2 synthetic blood; 1 vial of sample 3 synthetic blood; 1 vial of sample 4 synthetic blood; 1 vial of synthetic anti-D (anti-Rh) serum; 1 vial of synthetic anti-A serum; 1 vial of synthetic anti-B serum; 1 blood typing slide; 4 blue mixing sticks; 4 yellow mixing sticks; 4 white mixing sticks; paper towels; disposable gloves

Risks

WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
Staining from simulated blood	Simulated blood will stain skin and clothing. Avoid any direct contact with skin and clothing and wear appropriate personal protective equipment (PPE), such as gloves and lab coat.

What to do

1. Collect your blood typing slide and examine it. You will observe that the slide contains three wells, labelled A, B and Rh.
2. Collect sample 1 and add one drop of the synthetic blood sample to each well using the dropper vial. Replace the cap on the vial to prevent cross-contamination between different samples. Ensure you replace the lid each time before opening a new vial.
3. Add a single drop of each synthetic antibody serum to the appropriate well as listed in Table 1.

TABLE 1 Labelled wells and corresponding synthetic antibody serum

WELL ON BLOOD TYPING SLIDE	ANTIBODY SERUM
A	Anti-A (blue)
B	Anti-B (yellow)
Rh	Anti-Rh (white)

4. Using a mixing stick, carefully mix the synthetic blood and antibody serums for 30 seconds. Use a different mixing stick for each sample to avoid cross-contamination. Use a blue mixing stick for anti-A, yellow for anti-B and white for anti-Rh.
5. Once all the serums have been mixed into the synthetic blood sample, examine the samples in each well and look for the presence or absence of agglutination. If the liquid has a uniform texture, no agglutination has taken place. A granular sample will indicate that agglutination has taken place.
6. Record your results in Table 2 in the 'Sample 1' column. Simply write 'yes' or 'no' as to whether agglutination occurred with each antibody serum.
7. Rinse the blood typing slide thoroughly over the sink, using tap water. Dry with a paper towel.
8. Repeat the same procedure (steps 2–7) using synthetic blood samples 2, 3 and 4.
9. Once you have your results for each synthetic blood sample, use the results in Table 2 to determine the blood type of each sample. Remember, a reaction (agglutination) indicates that the synthetic blood sample contains the corresponding antigen to the antibody serum. Record your results in Table 3.

9780170449090

Studying your results

Copy and complete the tables below with the results of your experiment.

TABLE 2 Synthetic blood sample results

ANTIBODY SERUM	SAMPLE 1	SAMPLE 2	SAMPLE 3	SAMPLE 4
Anti-A				
Anti-B				
Rh				

TABLE 3 Blood type results

SAMPLE	BLOOD TYPE
Sample 1	
Sample 2	
Sample 3	
Sample 4	

Investigations

1. A break-in occurred at Harold's Jewellery Store on Friday night. The thief smashed all the windows and ran off with $20 000 worth of jewels. However, the thief was cut by some broken glass in their rush to escape the fast-approaching police. The forensic team collected a sample of the thief's blood that was left behind on a shard of broken glass. After conducting blood tests, the forensic team was able to identify the thief's blood type as O–. A witness saw somebody running away from the jewellery store shortly after the crime was committed and provided police with a description of their appearance. The police then brought in a suspect matching the description and noticed a cut on their leg. The police collected a sample of the suspect's blood to test for blood type. The suspect's blood is mixed with anti-A serum and the test results clearly showed the suspect's blood is not the same as the blood found at the scene of the crime. How did the test results indicate that the blood at the crime scene did not come from this individual?
2. Imagine another suspect was tested with anti-A or anti-B and their blood did not agglutinate, but when tested with anti-Rh it did. Does this information mean the suspect may have committed the crime? Explain your answer.
3. Jeff and Kim are first-time donors taking part in a Red Cross blood drive. Before they can donate, their blood needs to be typed. Jeff is A+. Kim is AB+.
   a. Identify which ABO antibody/ies are present in Jeff's blood.
   b. Identify which ABO antigen/s are present in Kim's blood.
4. Both Jeff and Kim's donated blood is sent to be processed and in both samples the blood cells are separated from the plasma. The separated cells and plasmas are then to be used in blood transfusions. But a blood researcher wishes to extract and identify the antigen A in Jeff's blood. Should the researcher attempt to extract the antigen A from his blood cells or his plasma?

9780170449090

CHAPTER 5 SUMMARY

- The circulatory system transports blood throughout the body to supply the needs of cells and remove their wastes.
- Blood is made up of plasma and formed elements (erythrocytes, leucocytes and thrombocytes).
- Plasma is water and dissolved substances. The most common mode of transport for carbon dioxide is in the plasma as bicarbonate ions (HCO_3^-).
- Erythrocytes, or red blood cells, contain haemoglobin, which is able to transport oxygen by forming oxyhaemoglobin.
- Leucocytes, or white blood cells, are important in protecting the body from infection.
- Thrombocytes, or platelets, are important in blood clotting.
- Blood clotting, or coagulation, allows a plug to form in a damaged vessel to restrict blood loss. Clots form with a mesh of fibrin which traps cells, platelets and plasma. The clot gradually retracts, becoming denser and stronger.
- The heart is a four-chambered pump that pushes blood around the body.
- Blood moves through the structures in this order: venae cavae, right atrium, right ventricle, pulmonary artery, lung, pulmonary vein, left atrium, left ventricle, aorta, body.
- The heart muscle contracts during systole and relaxes during diastole. The atria contract, then the ventricles.
- Valves prevent the blood flowing backwards.
- Blood vessels – arteries, capillaries and veins – transport blood from and to the heart.
- Arteries take blood away from the heart, while veins take blood towards the heart.
- Capillaries join the arteries and veins.
- Arteries contain blood under pressure. Their walls contain muscle and elastic fibres to maintain the pressure and allow vasoconstriction and vasodilation.
- Veins contain blood under low pressure. Their walls are thinner than arteries' walls, but they contain valves to stop the blood flowing backwards.
- Capillaries form a network through the cells. Their walls are only one cell thick, allowing substances to pass between the blood and cells.
- Red blood cells contain antigens on their surface.
- The ABO blood groups are due to sugar antigens: antigen A and antigen B.
- The body recognises the antigens that are found on its own cells, but it will produce antibodies for any antigens that are not normally found in the body.
- Rh blood groups are due to protein antigens. A person who is Rh positive has the antigen and someone who is Rh negative does not have the antigen. If an Rh-negative person receives Rh-positive blood, they will produce antibodies for the antigen.
- It is important that the blood groups are matched for blood transfusion so that the person receiving the blood will not produce antibodies that will react to the donated blood.
- There are different types of blood transfusions: whole blood, red cell concentrates, plasma, platelet concentrates, cryoprecipitate and autologous transfusion.
- The lymphatic system collects fluid that leaks from blood vessels and returns it to the blood, as well as playing a role in defending against disease.
- Lymph vessels are blind-ended tubes that collect fluid, proteins and pathogens from between the cells.
- Fluid, called lymph, moves through the vessels due to the constriction of muscles. Valves stop the lymph moving backwards.
- Lymph nodes have a connective tissue framework with lymphocytes, macrophages and plasma cells.
- Lymph is filtered as it moves through the lymph nodes.

9780170449090

CHAPTER 5 GLOSSARY

ABO blood group system A method of classifying blood types according to the antigens on the surface of the red blood cells

Agglutination The clumping together of micro-organisms or of blood cells

Alveoli Air sacs in the lungs

Antibody A substance produced in response to a specific antigen; it combines with the antigen to neutralise or destroy it

Antigen Any substance capable of causing the formation of antibodies when introduced into the tissue

Arteriole A very small artery

Artery A blood vessel that carries blood away from the heart

Atria The top chambers of the heart; singular: atrium

Atrial systole Contraction of the atria of the heart

Atrioventricular valves Valves within the heart that ensure the blood flows through it in one direction only

Autologous transfusion A transfusion using the patient's own blood

Biconcave Shaped concave on both sides, dipping inwards in the centre

Blood clotting Formation of a blood clot; also known as coagulation

Capillary A microscopic blood vessel that links arterioles and venules

Carbaminohaemoglobin A molecule resulting from a combination of carbon dioxide and haemoglobin

Cardiac cycle The cycle of events that occurs in one complete heartbeat

Cardiac muscle The muscle that forms the wall of the heart

Cardiac output The volume of blood pumped from one ventricle of the heart in one minute

Chordae tendineae Tendon-like structures that connect papillary muscle to valves

Circulation The movement of blood through the heart and blood vessels

Circulatory system The body's transport system, consisting of the heart, blood, blood vessels, lymph and lymph vessels

Clot Blood cells, platelets and plasma trapped together in a mesh of fibrin

Clot retraction Contraction of the fibrous threads of a blood clot

Clotting factors Chemical substances in blood plasma that allow blood to clot

Coagulation The process of blood becoming gel-like; also called clotting

Cryoprecipitate A blood product used in transfusions; produced by freezing the plasma and thawing it slowly

Deoxygenated blood Blood that contains little oxygen

Diastole The period of relaxation of the heart (between contractions), during which it fills with blood

Erythrocyte *see* red blood cell

Fibrin An insoluble protein that forms blood clots by holding blood cells, platelets and plasma together in a mesh

Formed element Any cell or cell-like structure in the blood

Haematocrit The ratio of red blood cells to the total volume of blood

Haemoglobin The pigment in red blood cells; it is involved in the transport of oxygen and some carbon dioxide through the body

Haemophilia An inherited disorder in which the blood clots slowly or not at all

Heart A hollow, muscular organ that pumps blood

Heart rate The number of heartbeats per minute

Immunoglobulins A group of proteins; antibodies are immunoglobulins

Inferior vena cava A large vein carrying blood from the lower body to the right atrium

Intercellular fluid Fluid between the cells

Leucocyte/leukocyte A white blood cell

9780170449090

Lymph Colourless fluid that circulates through the lymphatic vessels before returning to the blood

Lymph gland *see* lymph node

Lymph node An oval or bean-shaped structure found on the lymphatic vessels; it is involved in protection against infection; also called a lymph gland

Lymph vessels *see* lymphatic vessel

Lymphatic system A system of vessels that drains excess fluid from the tissues; also called the lymph system

Lymphatic vessel A large vessel that collects lymph from the lymph capillaries; lymphatic vessels join up and eventually return lymph to the blood

Lymphocyte A type of white blood cell; also found in lymph nodes and in lymph

Lymphoid tissue Tissue containing many lymphocytes and macrophages; found mostly in the lymph nodes but also in the bone marrow, tonsils, spleen and thymus

Macrophage A phagocytic cell derived from a monocyte (a type of white blood cell)

Metabolic wastes Substances produced by cells that cannot be used and that would be harmful if allowed to accumulate

Oxygenated blood Blood containing a lot of oxygen

Oxyhaemoglobin Oxygen combined with haemoglobin

Papillary muscles Muscles in the ventricles of the heart that anchor the valves

Pericardium Membrane enclosing the heart

Phagocytic cell Cell that can engulf and digest micro-organisms and cell debris

Plasma The fluid part of the blood in which the cells are suspended

Plasma cell Cell that develops from a B cell and produces antibodies

Platelet One of the formed elements of blood; a fragment of cytoplasm enclosed in a membrane but lacking a nucleus; also called a thrombocyte

Platelet concentrate A component of blood used in transfusions

Pulmonary vein The vein that transports blood from the lungs to the left atrium

Red blood cell One of the formed elements of the blood; contains haemoglobin

Red cell concentrate A component of blood used in transfusions; produced by spinning blood in a centrifuge

Rh blood group system A method of classifying blood types according to the antigens on the surface of the red blood cells

Semilunar valve Valve preventing blood from flowing back into the ventricles; located at the start of the aorta and pulmonary artery

Septum The partition between the left and right sides of the heart

Serum The protein-rich fluid that separates out when blood coagulates

Sternum The breastbone

Stroke volume The volume of blood pumped from the left ventricle during one contraction

Superior vena cava The large vein taking blood from the top of the body to the right atrium

Systole The period when heart muscle contracts

Thrombocyte *see* platelet

Thrombus *see* clot

Transfusion The transfer of blood, or of some of the components of blood, into the circulation of a person

Vasoconstriction Decrease in the diameter of blood vessels, restricting the flow of blood

Vasodilation Increase in the diameter of blood vessels, increasing the flow of blood

Vasodilator Substance that produces a local widening, or dilation, of blood vessels

Vein A blood vessel that carries blood towards the heart

Ventricles The bottom chambers of the heart

Ventricular systole The phase of the heartbeat when the ventricles contract

Venule A small vein

Wastes *see* metabolic wastes

White blood cell One of the blood cells; it contains a nucleus, but no haemoglobin

Whole blood Blood taken from a donor, with a chemical added to prevent clotting; used in transfusions

9780170449090

CHAPTER 5 REVIEW QUESTIONS

Recall

1 Describe the external appearance of the heart.
2 Draw a simple diagram of the human heart and show the direction of blood flow.
3 State the function of valves in the circulatory system.
4 List the functions of blood and explain the importance of each function.
5 a Describe the ways in which oxygen is carried in the blood.
b Describe the ways in which carbon dioxide is carried in the blood.
6 Define 'circulation'.
7 Describe the two ways in which blood flow to an organ can be increased.
8 Briefly describe the structure of a lymph node.
9 Describe the sequence of events that occurs in blood clotting.
10 List the antigens and antibodies involved in the ABO blood group system.

Explain

11 After passing through the capillaries of the body, the blood returns to the heart to be pumped again before going through the lungs. Explain why the blood must be pumped twice for each complete circulation through the body and lungs.
12 a Why are valves necessary in the heart?
b Explain how the atrioventricular valves work.
c Explain how the semilunar valves work.
13 Explain the changes in blood flow that occur during exercise.
14 For each of the following, explain how its structure is related to its function.
a Red blood cell
b Artery
c Heart
15 Explain how the lymphatic system achieves its function.
16 Discuss the differences between the lymphatic and circulatory systems.
17 Explain why early attempts at blood transfusion were sometimes successful but, more often, led to the death of the patient. What would have caused those patients to die?

Apply

18 Why is blood red?
19 Explain why someone with an infected toe may experience a lump in their groin.
20 If the heart contracts 70 times per minute, how many times does it contract in a day (24 hours), and how many times in a week?
21 When blood plasma is given in a transfusion, would the donor of the plasma have to be the same ABO blood group as the receiver? Explain your answer.

Extend

22 People living near sea level have about 5 million erythrocytes in 1 mm^3 of blood. Those living at an altitude of 5500 m above sea level have about 7.5 million erythrocytes per 1 mm^3 of blood. Suggest an explanation for this difference.
23 At one time, it was believed that disease was caused by 'bad blood'. Taking large amounts of blood from a patient by bleeding (blood-letting) was widely practised as a cure for disease. Louis XIII of France had blood taken 47 times in six months; Louis XV was bled 38 times,

9780170449090

and Charles II of England had blood taken numerous times, even just before his death. Describe some of the effects that the removal of large quantities of blood would have on a patient.

24 Which do you think is more important to the body – the heart or the capillaries? Explain your answer.

25 If you remain sitting for a long time, such as on a long flight, you may experience swelling of the feet and ankles. Suggest why this occurs.

26 Haemolytic disease of the newborn may occur if a mother has Rh-negative blood and her developing foetus has Rh-positive blood. Some of the foetus's blood may leak across the placenta and mix with the mother's blood. The mother will then produce anti-Rh antibodies, which can move back across the placenta and destroy the baby's red cells. The cause of haemolytic disease of the newborn was discovered in 1940. A vital clue that enabled scientists to determine the cause was that first babies were never affected by the condition unless the mother had previously received a transfusion of Rh-positive blood.

- **a** Why would first babies rarely be affected by this condition?
- **b** Why would the condition be more likely to occur if a mother had previously received a transfusion of Rh-positive blood?

27 The table below shows the result of testing blood samples from three different individuals with group A plasma and group B plasma. What is the blood group of each of the individuals D, E and F?

TESTED WITH	INDIVIDUAL		
	D	E	F
Group A serum	Clumping	No clumping	Clumping
Group B serum	No clumping	No clumping	Clumping

9780170449090

6 THE DIGESTIVE SYSTEM SUPPLIES NUTRIENTS FOR THE BODY

UNIT 1 CONTENT

SCIENCE INQUIRY SKILLS

» conduct investigations, including monitoring body functions; use microscopy techniques; and perform real or virtual dissection, safely, competently and methodically for the collection of valid and reliable data

SCIENCE UNDERSTANDING

Digestive system

» the supply of nutrients in a form that can be used in cells is facilitated by the structure and function of the digestive system at the cell, tissue and organ levels

» digestion involves the breakdown of large molecules to smaller ones by mechanical digestion (teeth, bile and peristalsis) and chemical digestion (by enzymes with distinctive operating conditions and functions that are located in different sections of the digestive system)

» the salivary glands, pancreas, liver and gall bladder produce or store secretions which aid the processes of digestion

» absorption requires nutrients to be in a form that can cross cell membranes into the blood or lymph and occurs at different locations, including the small intestine and large intestine

» elimination removes undigested materials and some metabolic wastes from the body

Source: School Curriculum and Standards Authority, Government of Western Australia

9780170449090

All cells require nutrients in order to provide energy for the cell's activities and materials for cell growth, cell reproduction, secretion and other metabolic processes. The **digestive system** extracts nutrients from the food we eat and absorbs them into the body for use by the cells.

The organs of the digestive system are structured and arranged so that they can carry out six basic activities:

1. ingestion of food and water
2. mechanical digestion of food
3. chemical digestion of food
4. movement of food along the alimentary canal
5. absorption of digested food and water into the blood and lymph
6. elimination of material that is not absorbed.

The structures of the digestive system work together to fulfil these functions.

6.1 TYPES OF DIGESTION

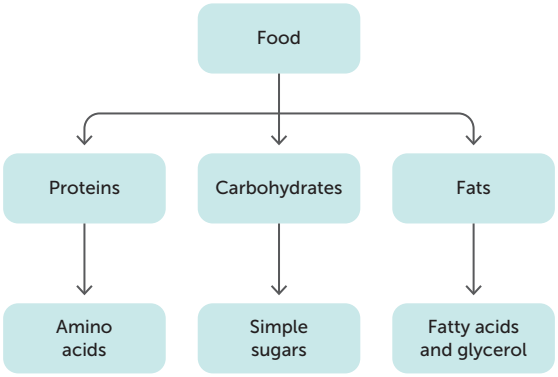


FIGURE 6.1 Macromolecules are made up of smaller units

Body cells require simple sugars, amino acids, fatty acids, vitamins, minerals and water to function normally. Vitamins, minerals and water are in the form of molecules that are small enough to be able to pass through the differentially permeable membrane surrounding each cell. Simple sugars, amino acids and fatty acids are eaten as complex carbohydrates, proteins and fats. The molecules of these substances are large and must be broken into smaller units before they can be absorbed into cells. The process in which carbohydrate, protein and fat molecules are broken down to products small enough to be absorbed into the blood and into the cells is called **digestion**.

Key concept

Digestion is the process of breaking down food into particles small enough to be absorbed into the bloodstream.

Mechanical digestion

Mechanical digestion is the physical breakdown of food particles. It involves the following processes in the mouth, stomach and small intestine:

- The teeth cut, tear and grind the food.
- Churning action in the stomach breaks the food down further.
- The gall bladder releases bile into the small intestine. Bile salts act as emulsifying agents, breaking fat down into smaller droplets.

The aim of mechanical digestion is to break the food down into smaller pieces so that the total surface area increases. This allows more effective chemical digestion, as the chemicals can access more of the food.

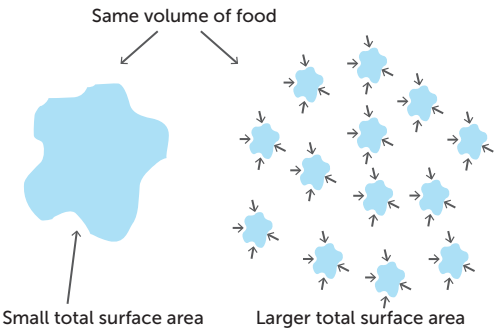


FIGURE 6.2 Mechanical digestion increases the total surface area, which speeds up digestion

Key concept

Mechanical digestion is the physical breakdown of food into smaller pieces to increase the surface area.

Chemical digestion

During **chemical digestion**, chemicals break down large, complex molecules into smaller, simpler molecules. These smaller molecules are then small enough to be absorbed into the bloodstream.

- Carbohydrates split into monosaccharides such as glucose, fructose and galactose.
- Proteins are split into peptides and amino acids.
- Lipids are split into fatty acids and glycerol.
- Nucleic acids are split into nucleotides.

Chemical digestion is achieved by enzymes. Enzymes are biological catalysts – chemicals that are able to increase the rate of a reaction without being consumed. To learn more about enzymes and how they work, refer to Chapter 3.

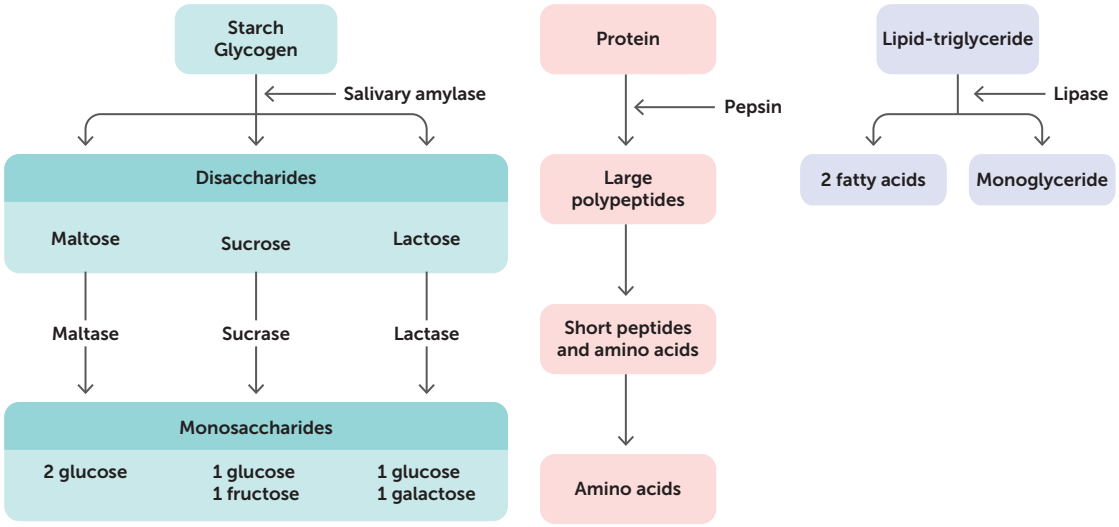


FIGURE 6.3 Chemical digestion uses enzymes to break complex molecules into simpler molecules

Key concept

Chemical digestion uses enzymes to break large, complex molecules into small, simpler molecules.

Questions 6.1

RECALL KNOWLEDGE

1 List the types of digestion.

2 Complete the following table.

LARGE, COMPLEX MOLECULE	SMALL, SIMPLE MOLECULE
Protein	
	Monosaccharides
Triglycerides (lipids)	

3 Describe the purpose of digestion.

APPLY KNOWLEDGE

4 List two similarities and two differences between mechanical and chemical digestion.

5 People who have had their gall bladder removed are unable to control the release of bile into the small intestine. Predict a consequence of this.

9780170449090

6.2 THE ALIMENTARY CANAL

The **alimentary canal** is the continuous tube that runs from the mouth to the anus. Together with associated organs such as the pancreas and the gall bladder, the alimentary canal makes up the digestive system. The lining of the alimentary canal is the surface through which nutrients are absorbed.

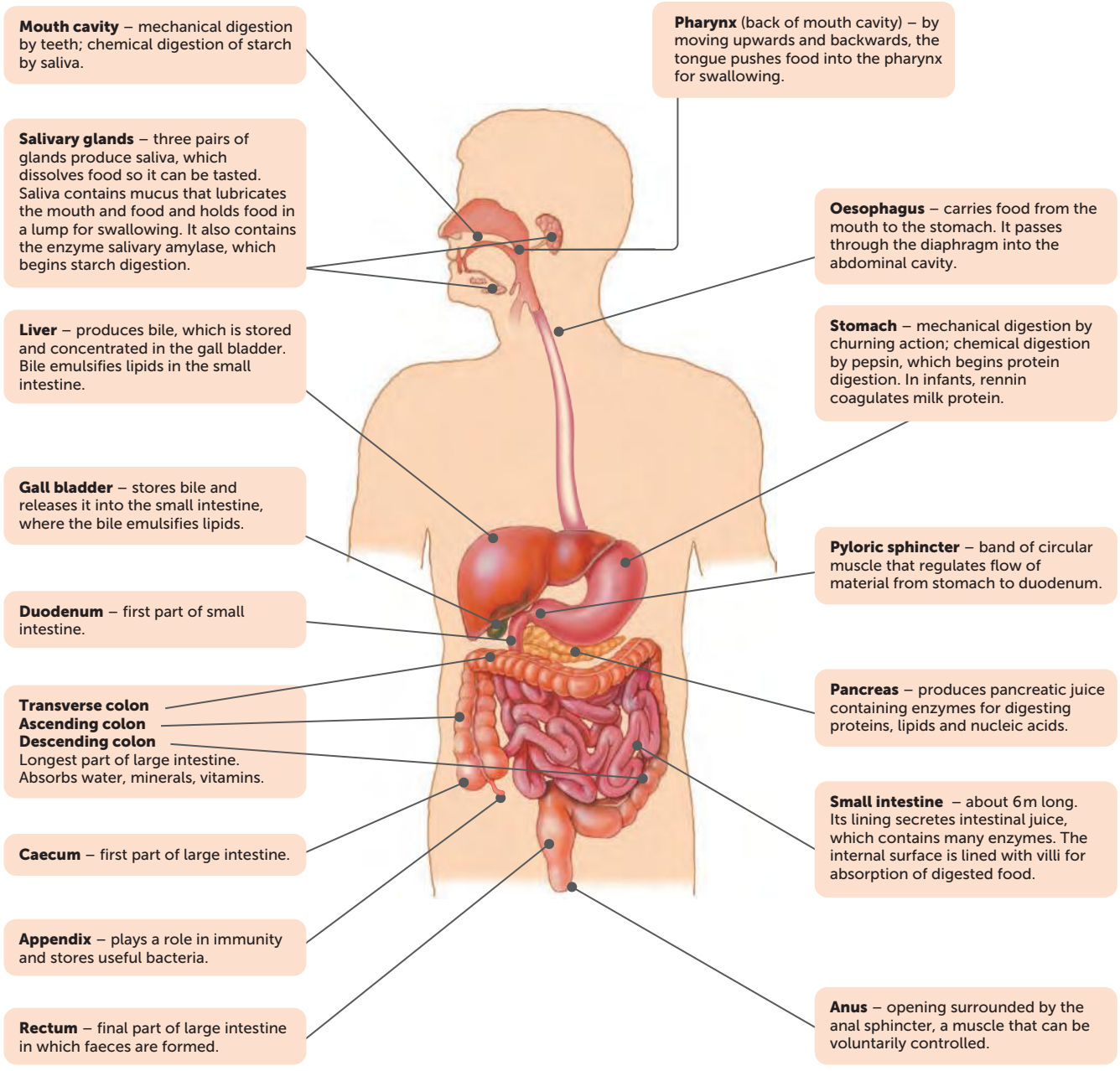


FIGURE 6.4 Structure and functions of parts of the digestive system

9780170449090

The mouth

Intake of food, called **ingestion**, occurs at the mouth. Here the food is chewed in a process called **mastication**. Both mechanical and chemical digestion commence before the food is swallowed.

Activity 6.1
Investigating amylase metabolism

Saliva and chemical digestion

As the food is chewed it is mixed with **saliva**, a fluid that is secreted into the mouth cavity by three pairs of **salivary glands**. It contains mucus to lubricate the food and a digestive enzyme – salivary amylase – which begins the chemical digestion of starch into the disaccharide maltose.

The teeth and mechanical digestion

The action of the jaws and teeth begins mechanical digestion. There are four types of teeth, each with a different function in mechanical digestion. A full adult set of teeth in the lower jaw consists of:

- four **incisors** – chisel-shaped teeth used for biting or cutting, as when taking a bite of an apple
- two **canines**, one on each side of the incisors. These are conical teeth used for tearing. Human canines are the same length as the other teeth
- four **premolars**, two on each side of the jaw
- six **molars**, three on each side of the jaw. The premolars and molars have broad crowns with rounded cusps. The cusps of the teeth of one jaw fit into depressions on the surface of teeth on the other jaw, making the premolars and molars ideal for crushing and grinding food.

The same number and type of teeth occur in the upper jaw.

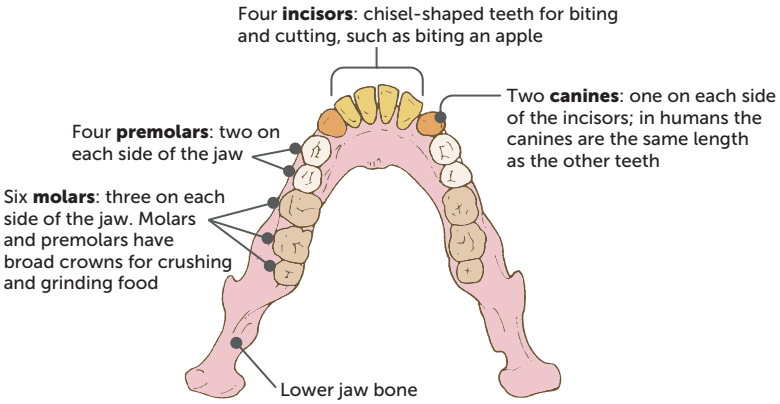


FIGURE 6.5 Adult teeth in the lower jaw

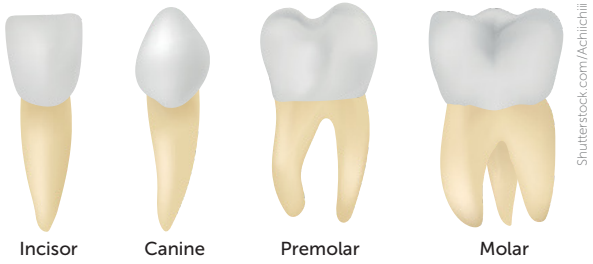


FIGURE 6.6 The shape of different types of teeth

After chewing, the tongue shapes the food into a rounded lump called a **bolus**. To swallow, the tongue moves upwards and backwards, pushing the bolus into the back of the mouth, the **pharynx**, which leads to the oesophagus.

The oesophagus

The **oesaphagus** is a tube about 23–25 cm long that connects the pharynx to the stomach. The wall of the oesophagus, like the rest of the alimentary canal, has a double layer of muscle. **Circular muscle** has muscle fibres arranged in a circle, and **longitudinal muscle** has fibres arranged along the length of the canal.

9780170449090

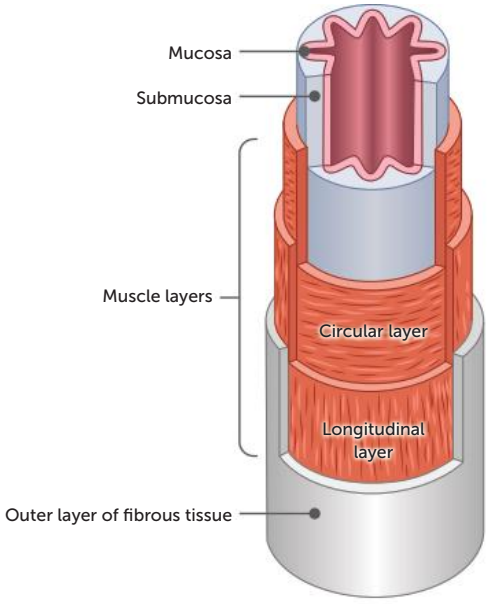


FIGURE 6.7 Structure of the oesophagus

Peristalsis
This website shows peristalsis in action.

Swallowing and peristalsis
This website shows swallowing and peristalsis.

As the lump of food enters the pharynx and oesophagus, the circular muscle behind it contracts to narrow the tube. The contraction of successive bands of circular muscle causes the constriction to move in a wave called **peristalsis**. This movement pushes the food in front of it, assisted by the secretion of mucus that lubricates the inner lining.

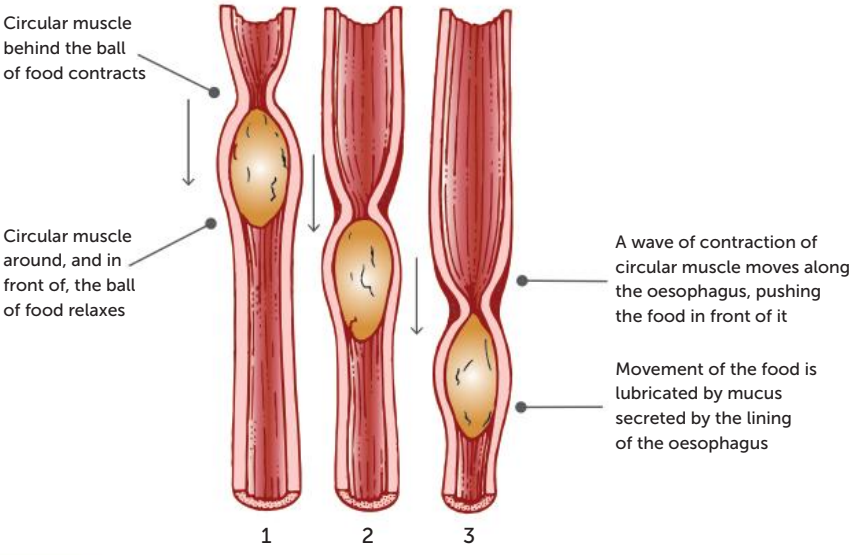


FIGURE 6.8 Food is moved along the oesophagus and along the rest of the alimentary canal by peristalsis

Key concept

Peristalsis is the wave-like muscle contractions that move food through a tube such as the oesophagus.

The stomach

The oesophagus passes through the diaphragm, a sheet of muscle that separates the thoracic cavity from the abdominal cavity. Just after passing through the diaphragm it opens into the **stomach**, a roughly J-shaped, enlarged section of the alimentary canal.

Mechanical digestion in the stomach is achieved by waves of muscular contraction that move along the stomach wall. Unlike the rest of the alimentary canal, the stomach has an oblique muscle layer as well as a circular and longitudinal layer. This enables the stomach to contract in a variety of ways to churn the food and mix it with the stomach juices until the food is converted to a thick, soupy liquid called **chyme**.

9780170449090

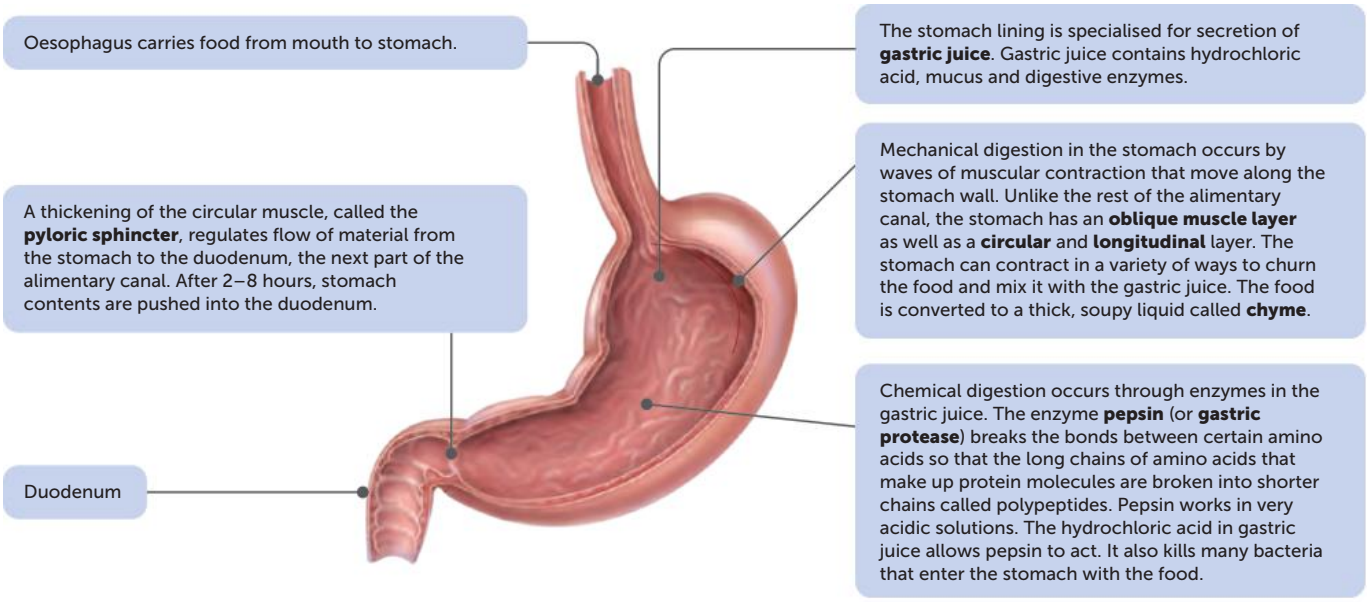


FIGURE 6.9 The stomach and its functions

The lining of the stomach, the **mucosa**, is specialised for the secretion of gastric juice by **gastric glands** located in narrow, tube-like structures called gastric pits. **Gastric juice** is a digestive juice containing hydrochloric acid, mucus and digestive enzymes. Each of these is secreted by a different type of cell in the gastric pits. Gastric juice is responsible for chemical digestion in the stomach, which is mainly the start of protein digestion.

The pH in the stomach is approximately 2–3, due to the hydrochloric acid. The cells lining the stomach are protected from the acid by a layer of mucus. The acidic environment allows the enzyme pepsinogen to be converted to pepsin, an active form of the enzyme. Pepsin is able to break proteins down into shorter peptides. It also breaks down the nucleic acids DNA and RNA.

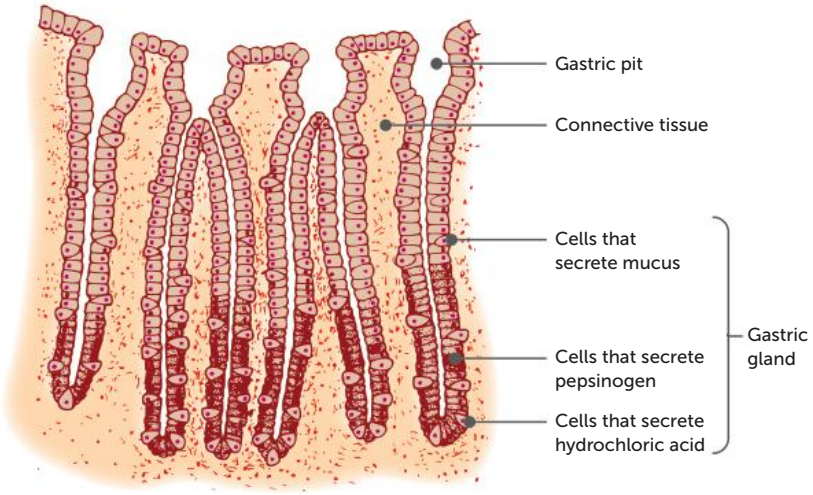


FIGURE 6.10 Mucosa of the stomach as seen in a transverse section

Nutrients are not absorbed into the bloodstream through the stomach, because the internal surface is covered by a thick layer of mucus. However, some alcohol and a few other drugs such as aspirin are absorbed.

At the lower end of the stomach there is a thickening of the circular muscle, which results in a constriction called the **pyloric sphincter**. The constriction is sufficient to prevent the stomach contents moving through unless pushed along by peristalsis. After 2 to 8 hours, the stomach contents are gradually pushed into the next part of the alimentary canal, the small intestine.

9780170449090

Key concept

Food undergoes mechanical and chemical digestion in the stomach.

The small intestine

The **small intestine** gets its name from the narrow diameter of its tube. It is the longest part of the alimentary canal, at approximately 6–7 m in length. It receives material pushed through the pyloric sphincter from the stomach.

There are three regions of the small intestine:

1. *Duodenum:* the first part of the small intestine. It is the shortest section at only 25 cm. The duodenum extends from the bottom end of the stomach in a curve around the pancreas. Most of the chemical digestion occurs here before the chyme moves further along the small intestine.
2. *Jejunum:* the middle section of the small intestine. The lining of the jejunum allows effective absorption of carbohydrates and proteins.
3. *Ileum:* the final part of the small intestine. It is here that vitamin B12, bile salts, and any remaining products of digestion are absorbed.

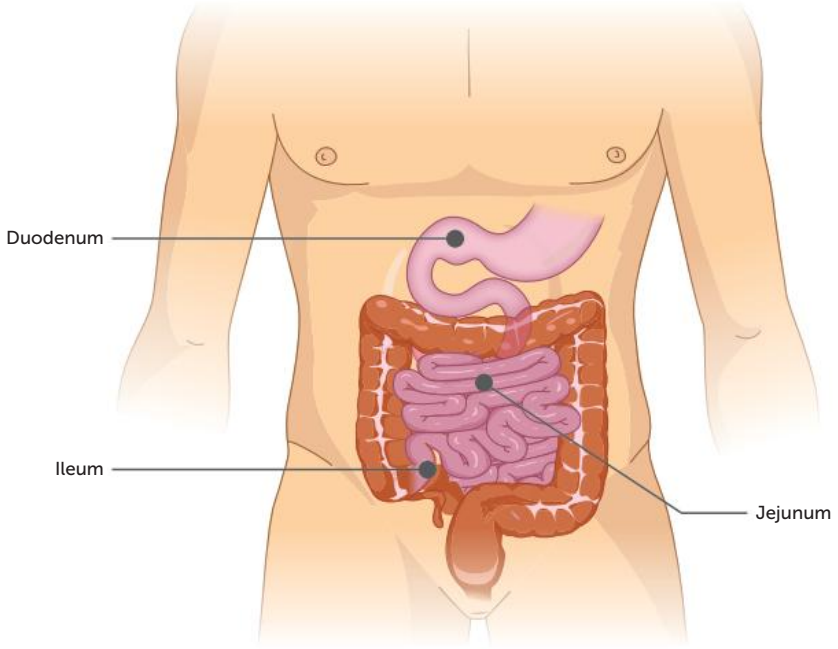


FIGURE 6.11 Parts of the small intestine

Digestion continues in the small intestine under the influence of:

- *pancreatic juice* – secreted by the pancreas via the pancreatic duct
- *bile* – produced by the liver, but stored in the gall bladder and secreted into the small intestine via the bile duct
- *intestinal juice* – secreted by glands in the lining of the small intestine.

Pancreatic juice

Pancreatic juice enters the duodenum through the common bile duct. It helps to neutralise the acid that has come with the material from the stomach and contains many of the enzymes involved in the digestion of food. The enzymes include:

- **pancreatic amylase,** which breaks down starch into the disaccharide maltose
- **trypsin** (or pancreatic protease), which splits proteins into peptides
- **pancreatic lipases** – enzymes that break down fats into fatty acids and glycerol
- **ribonuclease** and **deoxyribonuclease** – enzymes that digest RNA and DNA.

9780170449090

Bile

Bile is secreted into the small intestine through the common bile duct. Although it does not contain any digestive enzymes, **bile salts** are very important in the mechanical digestion of fats. They act like a detergent and **emulsify** the fat, breaking it into tiny droplets. This is a form of mechanical digestion, as it increases the surface area on which the lipases can act to bring about the chemical digestion of fat.

Intestinal juice

Intestinal juice contains many enzymes that complete the digestion of carbohydrates, proteins and lipids. These include:

- peptidase to break down peptides into amino acids
- sucrase, lactase and maltase to break down sucrose, lactose and maltose, respectively, into the monosaccharides glucose, fructose and galactose
- lipases to break down lipids into fatty acids and glycerol. (See chemical digestion in the small intestine in Table 6.1 on page 153.)

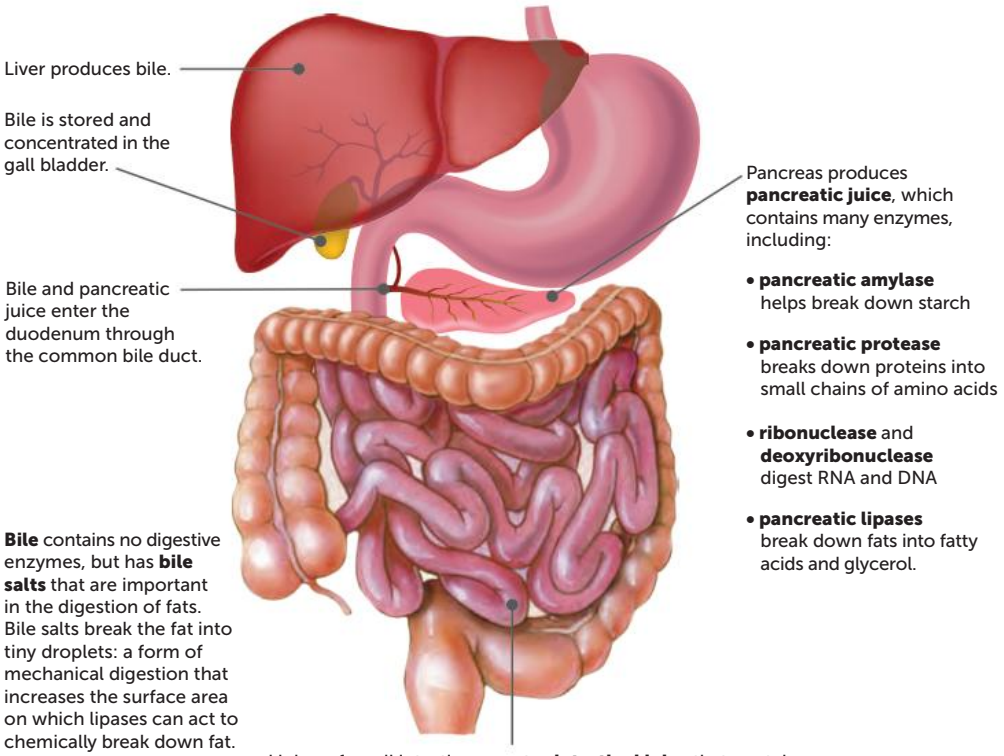


FIGURE 6.12
Digestion in the small intestine

Mechanical digestion also continues in the small intestine through a process called **segmentation**. Contractions of the circular muscles narrow the intestine which helps break up the bolus and mix it with the juices and bile.

Key concept

Enzymes in pancreatic juice and intestinal juice facilitate chemical digestion, while bile salts emulsify fat droplets.

9780170449090

Absorption of nutrients

When chemical digestion is complete, the complex carbohydrates will have been broken down into simple sugars, the proteins into amino acids, and the fats into fatty acids and glycerol. These products of digestion, along with substances such as vitamins, minerals and water, are then absorbed through the wall of the small intestine into the blood. Nutrients are absorbed through the internal surface of the small intestine, so efficient absorption requires a large surface area. A large internal surface area is achieved in a number of ways:

- The small intestine is very long – about 6–7 m.
- The inner lining, known as the **mucosa**, has folds that extend into the interior of the small intestine.
- The mucosa has small, finger-like projections called **villi** that extend from the folded surface.
- The cells covering the outside of the villi have tiny microscopic projections from their external surfaces. These are the **microvilli**.

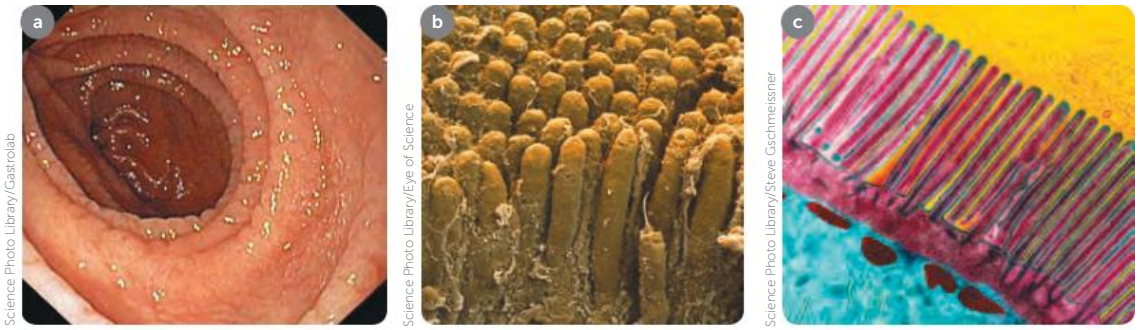


FIGURE 6.13 a Folds of the mucosa on the inside of the small intestine; **b** Scanning electron micrograph of villi that cover the internal surface of the small intestine; **c** Electron micrograph showing microvilli that cover the surface of each villus

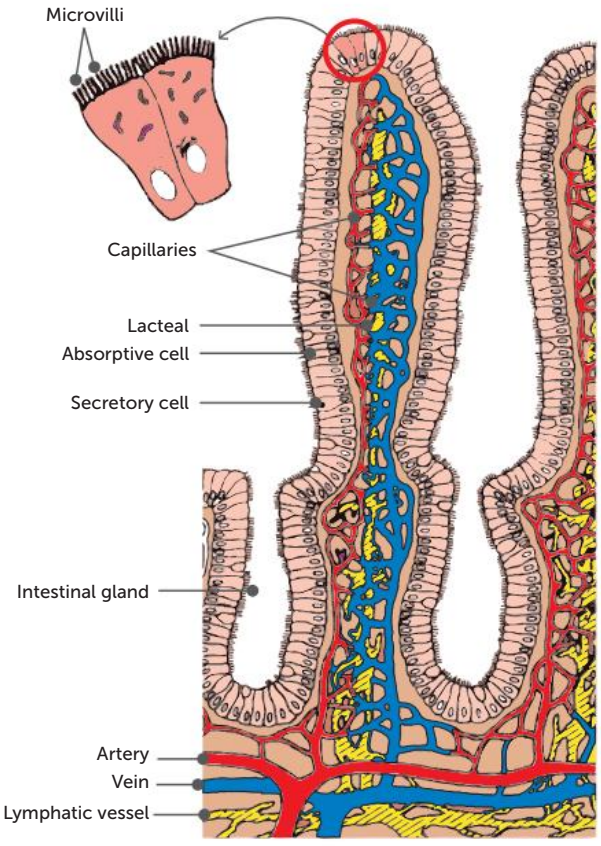


FIGURE 6.14
Structure of a villus

The structure of a villus is ideally suited to its function of nutrient absorption. Each villus is about 1 mm long, although the villi in the jejunum are longer than those in the duodenum and ileum. Each villus is covered by a single layer of cells. Inside the villus is a lymph capillary, called a **lacteal**, which is surrounded by a network of blood capillaries. Absorption is further enhanced by continual movement of the villi brought about by the muscular movements of the intestinal wall. This constantly brings the villi into contact with different parts of the intestinal contents. These contents are constantly changing as new material is emptied into the small intestine from the stomach.

Some absorption occurs through simple diffusion, as there is a higher concentration of nutrient materials in the interior of the small intestine than in the cells lining the villi. Absorption also occurs through **active transport**, which involves the cells of the villi using energy to take in

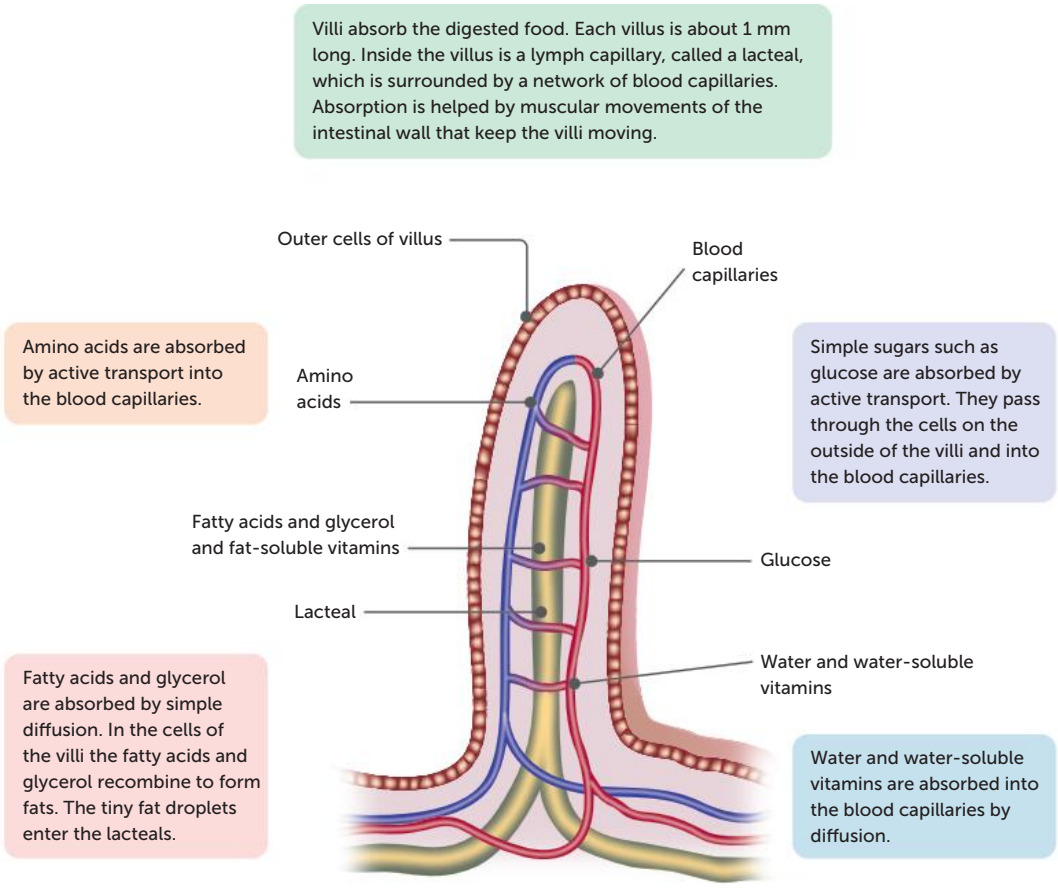


FIGURE 6.15 Absorption of nutrients into blood and lymph

nutrients against a concentration gradient – that is, taking in molecules from a lower concentration to a higher concentration.

From the walls of the villi, simple sugars, amino acids, water and water-soluble vitamins are absorbed into the blood capillaries. Fatty acids and glycerol recombine in the cells of the villi to form fats and, along with the fat-soluble vitamins, enter the lacteals. The substances that are absorbed into the blood capillaries are carried by the hepatic portal vein to the liver. Here they may be removed for further processing, or they may remain in the blood to be carried to other body cells. Substances that are absorbed into the lacteals are transported in the lymph system, which eventually empties into the blood through veins in the upper part of the chest.

Key concept

The lining of the small intestine has folds, villi and microvilli to maximise the absorption of nutrients.

The large intestine

The **large intestine** is about 1.5 m long, and is so named because it is larger in diameter than the small intestine. It is made up of the caecum, colon, rectum and anus. Additionally, the appendix attaches to the caecum.

There are no villi in the large intestine, and no digestive juices are secreted, although the lining does secrete a large amount of mucus. Movement of material through the large intestine is fairly slow, taking 18–24 hours for material to pass through. During this time, most of the remaining water is absorbed so the contents become more solid.

Bacteria in the large intestine break down much of the remaining organic compounds. Some bacteria produce vitamins, which are then absorbed through the walls into the blood. Mineral nutrients are also absorbed.

9780170449090

The semi-solid material left after water absorption and bacterial action makes up the faeces. **Faeces** contain water, undigested food material (particularly cellulose), bacteria, bile pigments (which give the faeces their colour) and the remains of cells that have broken away from the internal lining of the alimentary canal. The faeces pass through the rectum and anus to the exterior of the body.

Many people refer to defecation as 'excretion'. Excretion is the removal of metabolic waste – waste that has been produced by chemical activity of the body cells. Except for the bile pigments, the contents of faeces are not metabolic waste, so defecation is better referred to as **elimination**, rather than excretion.

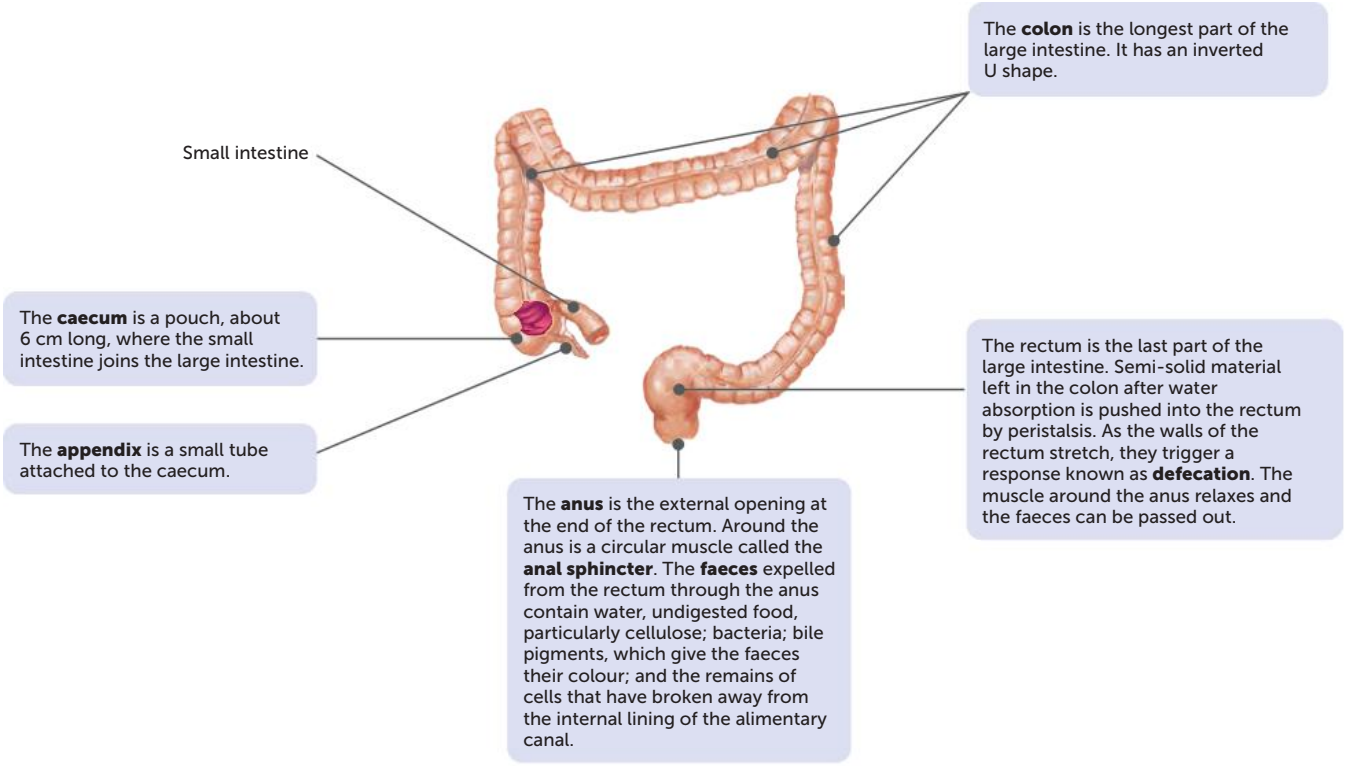


FIGURE 6.16 The large intestine structure and function

Key concept

The slow movement of material through the large intestine allows water to be absorbed, producing faeces.

WS

6.1 The digestive system

TABLE 6.1 Functions of parts of the digestive system

ORGAN	MECHANICAL DIGESTION	CHEMICAL DIGESTION	OTHER FUNCTIONS
Mouth	Breaks food into smaller particles by mastication	Saliva, which contains salivary amylase, begins starch digestion	Food is dissolved in saliva so that it can be tasted
Oesophagus		No chemical digestion	Transports food from the mouth to the stomach
Stomach	Waves of contraction churn food, producing chyme	Gastric juice contains the inactive pepsinogen. Hydrochloric acid will activate pepsinogen into pepsin, which breaks down proteins to polypeptides.	Stores large quantities of food as it is eaten; absorbs certain drugs, including some alcohol

TABLE 6.1 (*Continued*)

ORGAN	MECHANICAL DIGESTION	CHEMICAL DIGESTION	OTHER FUNCTIONS
Small intestine	Muscular contractions churn food; bile salts emulsify lipids	Pancreatic juice contains: • pancreatic amylase, which breaks starch into disaccharides • pancreatic protease, which breaks proteins and polypeptides into peptides • pancreatic lipases, which break lipids into fatty acids and glycerol • nucleases, which digest DNA and RNA. Intestinal juice contains: • amylases to break down disaccharides to simple sugars • peptidases to break down peptides to amino acids • lipases to break down lipids to fatty acids and glycerol	Absorbs simple sugars, amino acids, fatty acids, glycerol, vitamins, mineral nutrients and water through the villi
Large intestine	No chemical digestion		Absorbs water and vitamins; stores faeces; defecation

Activity 6.2
Investigating the action of pepsin

Activity 6.3
Investigating pancreatic juices

Questions 6.2

RECALL KNOWLEDGE

1 List the structures of the alimentary canal in order, starting from the mouth.
2 Which is longer: the oesophagus, small intestine or large intestine?
3 List the enzymes that are present in pancreatic juice.
4 Describe the function of the large intestine.
5 What type of digestion (mechanical or chemical) occurs in the stomach?

APPLY KNOWLEDGE

6 Explain how the lining of the small intestine maximises the absorption of nutrients.
7 Explain the role of hydrochloric acid in the stomach.
8 Herbivores, such as horses, have reduced canines and large premolars and molars. Explain the relevance of this.

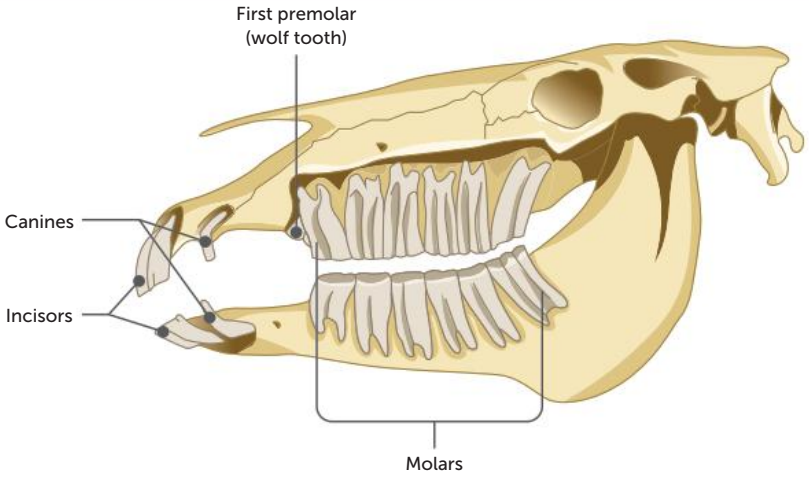


FIGURE 6.17 Diagram of a horse's teeth

9780170449090

6.3 THE EFFECT OF DIET ON THE ALIMENTARY CANAL

The speed with which material is moved through the alimentary canal depends on the size and contents of a meal. A large meal causes greater stretching of the stomach, and material is pushed into the small intestine much more quickly than when the stomach is less distended. High protein and/or high fat content in a meal slows the movement from stomach to small intestine. Alcohol and caffeine both stimulate movements of the stomach.

Constipation

Constipation occurs if the movements of the large intestine are reduced and the contents remain there for a long period of time. As water is absorbed, the faeces become drier and harder than usual. Defecation becomes difficult and possibly painful, a condition known as constipation. Constipation may be caused by a lack of roughage in the diet. Roughage is cellulose, or insoluble fibre, a major component of plant foods. Humans have no enzymes to digest cellulose, but it is important because it stimulates the movements of the alimentary canal. Other causes of constipation may be lack of exercise or emotional problems.

Diarrhoea

Diarrhoea is characterised by frequent defecation of watery faeces. It is caused by irritation of the small or large intestine, which increases peristalsis so that the contents of the intestines move through before there is adequate absorption of water. Such irritation may be the result of:

- a bacteria – for example, *Escherichia coli*, *Campylobacter*, *Salmonella* or *Vibrio cholerae*
- a virus – for example, the norovirus or rotavirus
- a parasite – for example, giardia
- cancer, such as bowel cancer
- coeliac disease
- lactose intolerance.

If the diarrhoea is severe, it may lead to dehydration through loss of water from the intestines, and may even cause death.

The importance of soluble fibre in the diet

Both soluble and insoluble fibre are found only in foods derived from plants. Soluble fibre includes pectins, gums and mucilage. Soluble fibre intake has been linked to lower cholesterol levels in the blood, decreased risk of heart disease and cancer, and beneficial effects on blood glucose levels. Fats in the intestines are trapped by soluble fibre, thereby helping to prevent their absorption by the body. This is thought to be the reason that soluble fibre helps to lower blood cholesterol levels. Good sources of soluble fibres are fruits, vegetables, oat bran, barley and soy products.

Bowel cancer

Bowel cancer, or **colorectal cancer**, is an uncontrolled growth of cells in the wall of the large intestine. Research suggests that bowel cancer may be linked to diet, high alcohol consumption and smoking. A diet high in red and processed meat, and low in fibre (fruit and vegetables), may increase the risk of developing bowel cancer. Being overweight or obese and physical inactivity are also risk factors.

9780170449090

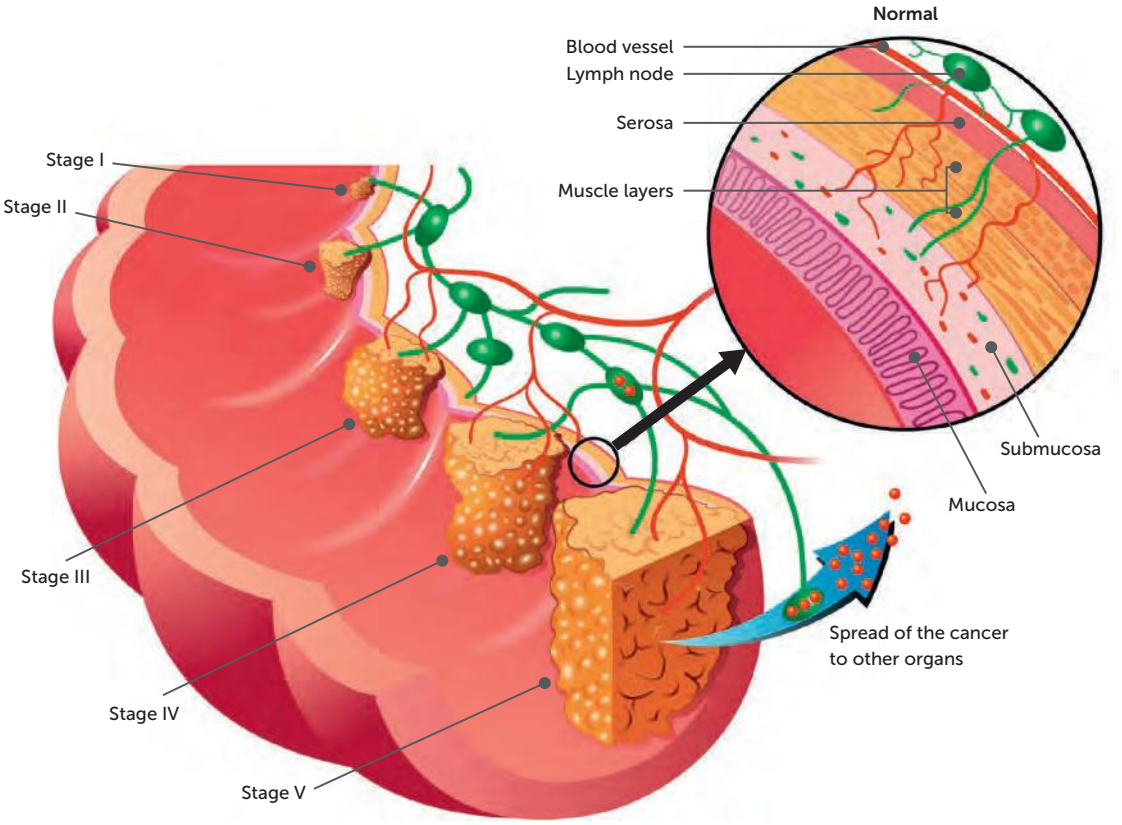


FIGURE 6.18 The different stages of bowel cancer

Coeliac disease

People with **coeliac disease** are unable to tolerate a protein called gluten, which is found in wheat, rye and barley. If such people eat food containing gluten, their immune system responds by damaging or destroying the villi in the small intestine. Without healthy villi, nutrients cannot be absorbed, and the person becomes malnourished no matter how much food they eat.

The symptoms of coeliac disease are many and vary from person to person. Symptoms such as muscle cramps, joint pain or tingling in the legs may appear to have nothing to do with nutrition or digestion. Some people may not have symptoms but are still in danger of becoming malnourished. The variety of symptoms makes the condition very difficult to diagnose.

Coeliac disease is inherited. There is no cure; the only treatment is to follow a gluten-free diet.

Key concept

A healthy diet is important for a healthy digestive system.

Questions 6.3

RECALL KNOWLEDGE

1 What is the common name for colorectal cancer?
2 List two conditions that are more likely with a diet low in fibre.
3 Define 'diarrhoea'.

APPLY KNOWLEDGE

4 Explain the difference between diarrhoea and constipation.
5 Identify the treatment for coeliac disease and justify its effectiveness.
6 Explain the relationship between peristalsis and diarrhoea.
7 Explain why it is important for people to eat fruit and vegetables.

9780170449090

CHAPTER 6 ACTIVITIES

SOUTHERN Biological Developed exclusively by Southern Biological

ACTIVITY 6.1 Investigating amylase metabolism

Functioning as biological catalysts, enzymes are proteins that increase the rate of chemical reactions and often play an essential role in digestion and metabolism. Amylase is an enzyme that breaks down starch into glucose and is responsible for giving food a sweet taste. Salivary amylase is produced by the salivary gland and can be found in the saliva of mammals. It is also produced by the pancreas and discharged into the small intestine. Salivary amylase functions to hydrolyse polysaccharides (carbohydrates) into disaccharides (sugars). Functioning optimally at the pH and temperature conditions of the human body, enzyme activity within amylase decreases or even ceases outside these conditions.

Aim

To determine the optimal temperature for amylase activity.
Time requirement: 45 minutes

You will need

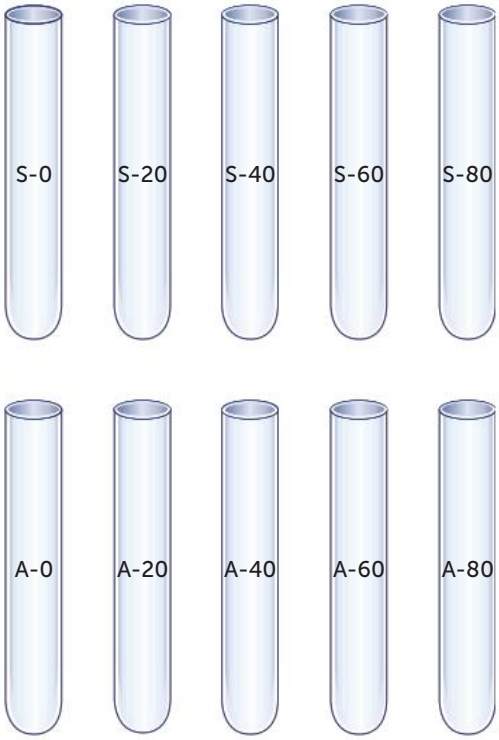
0.1 mol/L iodine solution; 1% amylase (clarase) solution (15 mL); 1% starch solution (15 mL); water baths; plastic pipettes; 10 test tubes; spotting tile (large enough for 40 drop tests); clock or timer; marker; thermometer; disposable gloves

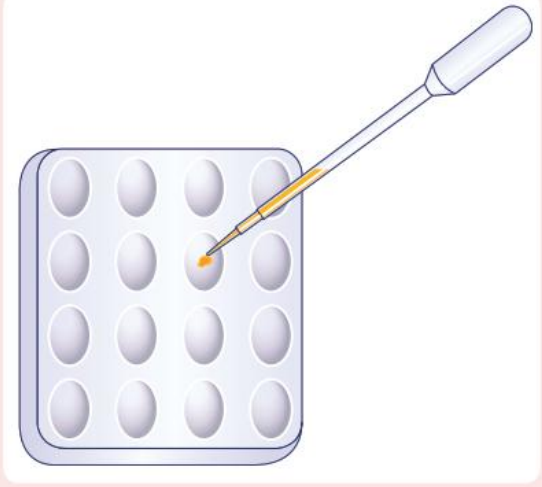
Risks

WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
Amylase can cause allergic or asthma symptoms if inhaled	Be aware of any allergies and avoid inhalation of aerosol droplets while handling solutions.
Iodine solution is an irritant	Avoid any contact with skin and eyes.
Disposable gloves may pose allergy risk	Use a type of glove that removes allergy risk and is suitable for the chemicals being used.
Working with high temperatures	To prevent scalding, take care when working with water baths with water temperatures higher than 50°C. Do not touch the outside of the glass beaker.

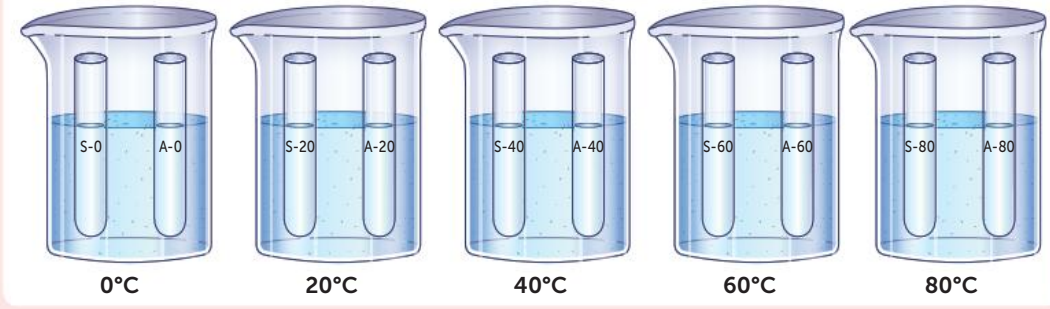
What to do

1. Prepare a series of water baths at 20°C temperature intervals (0°C, 20°C, 40°C, 60°C and 80°C).
2. Place a thermometer in each water bath to monitor temperature.
3. Collect 10 test tubes. Label them as shown in the figure opposite.
4. Using a pipette, add 2 mL of your 1% starch solution to the five 'S' test tubes.
5. Using a pipette, add 2 mL of your 1% amylase solution to the five 'A' test tubes.
6. Using a pipette, place a single drop of iodine into each dimple on a spotting tile. Ensure you have enough drops for 40 tests.
7. Using a pipette, place a sample of starch on to the first drop of iodine to act as your control, as this one will not change colour.





8 Place one starch test tube and one amylase test tube of each temperature variable into each water bath for five minutes.



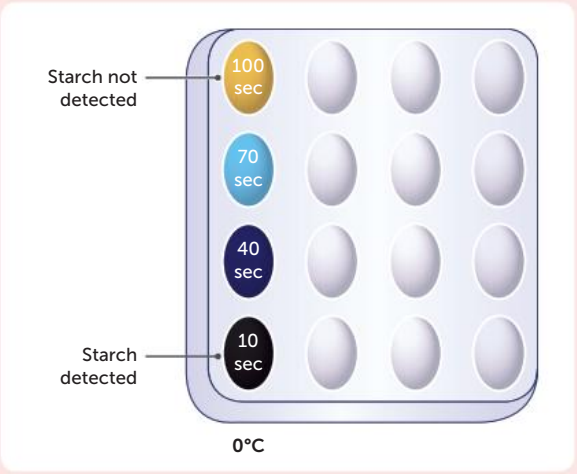
9 Remove the starch and amylase test tubes from the 0°C water bath and pour the contents of the amylase test tube into the starch test tube. Tap and lightly shake the test tube to mix the solutions. Place the test tubes back into the water bath and begin a timer.

10 Wait 10 seconds, then use a plastic pipette to remove a sample of the starch–amylase solution and transfer it to the spotting plate with a drop of iodine.

11 Continue taking samples from the test tube at 30-second intervals until the iodine returns to its original colour; this indicates the starch has been broken down. Ensure the test tube remains within the water bath to maintain the desired temperature.

12 Record the total time required for the solution to stop changing colour.

9780170449090



13 Repeat steps 9–11 for each of the temperature variables.
14 Record the results of your experiment in the table below. Create a graph using the collected data, indicating the total time taken for starch to be broken down at various temperatures.

Studying your results

1 Copy and complete the table with the results of your experiment.

TEMPERATURE (°C)	TIME TAKEN
0	
20	
40	
60	
80	

2 Graph your results to determine the following.
   a What was the optimal temperature for the amylase to break down starch?
   b Are your results consistent with the hypothesis (theory) that amylase works optimally at the temperature reflective of the human body?

Discussion

1 What is your independent variable?
2 What is the range of your independent variable?
3 What is your dependent variable?
4 Explain why the enzyme activity differed at 0°C compared to 100°C.
5 Why does the iodine solution change colour when the amylase–starch solution is added?
6 Why does the iodine solution stop changing colour as the experiment progresses?
7 Why are all the test tubes left in the water baths for five minutes before two solutions are added together?
8 Why was it important to keep the test tubes in the water bath at all times?
9 How accurate do you believe your collected data is? What may have influenced this accuracy? How would you modify the experiment to improve the level of accuracy?

Taking it further

Change the independent variable of this investigation and test the hypothesis that the amylase enzyme also works optimally at certain pH levels.

9780170449090

ACTIVITY 6.2 Investigating the action of pepsin

In this investigation, you will observe the action of pepsin on albumin, a globular protein. Pepsin is a digestive enzyme that is found in many organisms. It comes in many different forms, but in every case its function is to aid digestion by breaking proteins down via hydrolysis into their component amino acids.

Aim

To determine the optimal conditions for pepsin activity.
Time requirement: 45 minutes

You will need

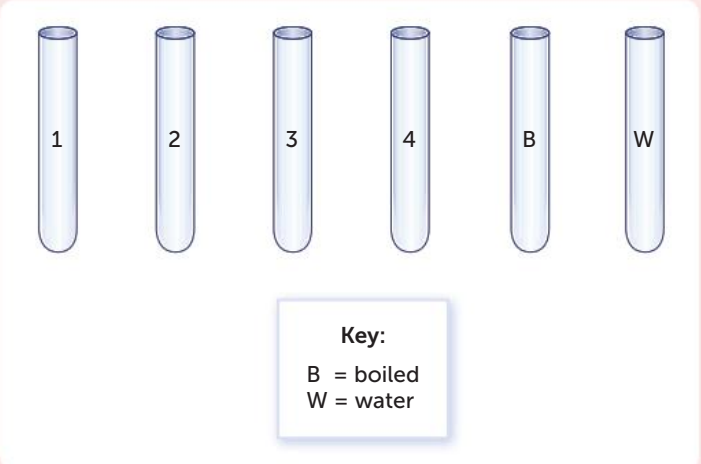
6 test tubes; test-tube rack; Bunsen burner; test-tube holder; plastic pipette; 0.1 mol/L hydrochloric acid solution; albumin suspension (20 mL); pepsin solution; 250 mL beaker; hot plate; marker; thermometer; distilled water; disposable gloves

Risks

WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
Bunsen burner flame can cause severe burns	Ensure safe use and avoid heating flammable liquids.
Albumin may cause allergic reactions	Consider restricting participation of students who have allergies to egg whites.
Disposable gloves may pose allergy risk	Use a type of glove that removes allergy risk and is suitable for the chemicals being used.
Working with high temperatures	To prevent scalding, take care when working with water baths with water temperatures higher than 50°C. Do not touch the outside of the glass beaker.

What to do

1 Collect six test tubes. Label them as shown.



2 Pour 5 mL of the albumin suspension into each of the four numbered test tubes.
3 Add 5 mL of distilled water to the tube labelled 'W'.

4 Add 2 mL of your pepsin solution to the tube labelled 'B' and bring to the boil over a Bunsen burner flame. The glass will become hot during this stage. To avoid burns, be careful and use the tube holder when handling the test tube.

5 Add three drops of distilled water to test tube 1.
6 Add three drops of dilute hydrochloric acid to the test tubes numbered 2, 3 and 4.
7 Prepare a water bath by filling a 250 mL beaker halfway with water and placing it on a hot plate. The water bath should be maintained at a temperature of approximately 40°C.
8 Place your test tubes in the water bath to warm them. It is only necessary to place the boiled test tube, labelled 'B', in the water bath if it has cooled down.
9 Add 1 mL of the warmed water in test tube W to test tube 2.
10 Add 1 mL of the boiled pepsin in test tube B to test tube 4.
11 Add 1 mL of warmed pepsin solution to test tubes 1 and 3.
12 Set your timer for 6 minutes. After 6 minutes, remove test tubes from the water bath and place them in the test-tube rack.
13 Observe the contents of the test tubes and compare their appearance. Record your results in the table below.

Studying your results

Copy and complete the table with the results of your experiment.

TEST TUBE	CONTENTS	RESULTS (DESCRIBE THE APPEARANCE)
1	Albumin, pepsin, water	
2	Albumin, water, HCl	
3	Albumin, pepsin, HCl	
4	Albumin, boiled pepsin, HCl	

9780170449090

Discussion

1. Why was the water added to test tubes 1 and 2 in those specific quantities?
2. Why is the albumin suspension cloudy? What is suggested by the clearance of the cloudiness?
3. What does the result in test tube 4 suggest?
4. What does the result in test tube 2 suggest?
5. Compare the results of test tubes 1 and 3. What can you infer from this comparison?
6. Antacids are bases that reduce the amount of acid in the stomach. This can reduce the discomfort associated with a highly acidic stomach environment. What might happen if more than the recommended number of antacids were consumed in a short period?
7. Can you think of another function of stomach acid besides digestion?

Taking it further

Test the effect of different antacid medications on gastric enzyme function.

SOUTHERN Biological Developed exclusively by Southern Biological

ACTIVITY 6.3 Investigating pancreatic juices

Pancreatic enzymes play a key role in digestion and nutrient absorption. Pancreatic juice is found in the small intestine and is a component of the fluid excreted by the pancreas. Trypsin and lipase are two enzymes that make up pancreatic juice. Trypsin functions to hydrolyse polypeptides, breaking down proteins into amino acids and producing water. Lipase breaks down lipids into glycerol and fatty acids, making them absorbable through the lining of the small intestine.

Aim

To observe polypeptides and lipid digestion by pancreatic enzymes.
Time requirement: 55 minutes

You will need

marker; two plastic test tubes; litmus milk, 20 mL; wooden stirrers; distilled or deionised water, 10 mL; pancreatin, 5% aqueous 10 mL; warm water bath (37–40°C); paper towels; clock or timer; plastic pipette; disposable gloves

Risks

WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
Pancreatin can cause allergic reactions in sensitive people	Be aware of any allergies.
Pancreatin can be irritating to the skin and eyes on contact	Wear appropriate personal protective equipment (PPE) at all times, including eye protection and gloves. Wash skin immediately if contact does occur.
Disposable gloves may pose allergy risk	Use a type of glove that removes allergy risk and is suitable for the chemicals being used.
Working with high temperatures	To prevent scalding, take care when working with water baths with water temperatures higher than 50°C. Do not touch the outside of the glass beaker.

- Wear appropriate PPE.
- Know and follow all regulatory guidelines for the disposal of laboratory wastes.
- Wash hands thoroughly before and after handling any chemicals.
- Sterilise work surfaces before and after the practical.
- Under no circumstances are the materials used in this practical to be consumed as food.

What to do

1. Collect two test tubes and label the tubes 1 and 2 using a permanent marker.
2. Using a pipette, add 10 mL of litmus milk solution to each test tube.
3. Add 5 mL of pancreatin solution to test tube 1. Using a clean stirrer, gently stir the contents of the test tube for one minute.
4. Add 5 mL of water to test tube 2. Ensure that you are adding the same volume of liquid to test tube 2 as you added to test tube 1.
5. Using a new, clean wooden stirrer, gently mix the water and litmus milk solution until fully dissolved.
6. Record the colour of the contents of each test tube.
7. Allow both test tubes to rest in a 37–40°C warm water bath for 15 minutes.
8. Carefully take the test tubes out of the water bath and observe the colour of the contents. Be careful not to splash hot liquid onto your skin or eyes. Record the colour of the contents of each test tube in the table below.

Studying your results

1. Copy and complete the table with the results of your experiment.

TUBE	CONTENTS	COLOUR BEFORE	COLOUR AFTER
1	Litmus milk, pancreatin powder		
2	Litmus milk, water		

2. Which solution indicated a positive result for acid production?

Discussion

1. What is the control in this experiment?
2. In which test tube were proteins and lipids digested? How do you know?
3. Explain the process behind the change in pH.
4. What role did trypsin play in this procedure?
5. What role did lipase play in this procedure?
6. Why did you have to place the test tubes in a water bath at 37°C? What is your hypothesis if this temperature was increased to 80°C? Decreased to 5°C?

9780170449090

CHAPTER 6 SUMMARY

- The digestive system extracts nutrients from the food we eat and absorbs them into the body for use by the cells.
- Digestion is the process of food being broken down into products small enough to be absorbed into the blood and cells.
- Mechanical digestion is the physical breakdown of food by cutting, tearing and grinding by the teeth, stomach and bile.
- Chemical digestion is the process of enzymes breaking large molecules into smaller, simpler molecules.
- Food is ingested in the mouth and mixed with saliva, where salivary amylase starts to break down starch.
- The different types of teeth – incisors, canines, premolars and molars – are each shaped to perform a specific function in mechanical digestion.
- Muscles in the oesophagus move the food from the pharynx to the stomach by the process of peristalsis.
- In the stomach, food is mixed with gastric juices to form chyme.
- Gastric juice, secreted from gastric glands, contains hydrochloric acid, mucus and enzymes to facilitate chemical digestion.
- The small intestine is 6–7 m long and is made up of the duodenum, jejunum and ileum, where digestion occurs due to intestinal juice, pancreatic juice and bile.
- Pancreatic juice contains pancreatic amylase, trypsin, ribonuclease, deoxyribonuclease and pancreatic lipase.
- Bile contains bile salts that emulsify fats.
- Intestinal juices contain amylases, peptidases and lipases.
- The lining of the small intestine is folded into villi, which are covered by microvilli. This results in a very large surface area for absorption.
- Absorption occurs through simple diffusion and active transport.
- The large intestine is 1.5 m long and is made up of the caecum, colon, rectum and appendix.
- Material moves slowly through the large intestine, allowing water to be removed and the formation of faeces.
- Bacteria in the large intestine break down organic compounds, producing vitamins.
- Diet can affect the digestive system, resulting in conditions such as constipation, diarrhoea, bowel cancer and coeliac disease.

9780170449090

CHAPTER 6 GLOSSARY

Active transport The use of energy to move substances, usually ions, across a cell membrane

Alimentary canal The tube via which food passes through the body, consisting of the mouth, oesophagus, stomach and intestines; also called the digestive tract

Bile A secretion of the liver, stored in the gall bladder and released into the small intestine

Bile salts Substances that break fats into tiny droplets

Bolus A ball-like structure of food and saliva

Canine The pointed tooth between the incisors and premolars

Chemical digestion The breakdown of food to small molecules by chemicals

Chyme The semifluid mass of partially digested food that leaves the stomach

Circular muscle Smooth muscle with fibres arranged in a circle around an organ, such as the stomach

Coeliac disease An autoimmune disease due to the immune system reacting to gluten

Colorectal cancer Cancer in the colon and rectum; also called bowel cancer

Constipation A condition in which defecation is difficult, with faeces being hard and dry

Deoxyribonuclease An enzyme in pancreatic juice that digests DNA

Diarrhoea The frequent passing of watery faeces

Digestion The mechanical and chemical breakdown of food to small molecules that can be absorbed into the body

Digestive system The system that breaks down the food taken into the body ready for absorption into the cells

Elimination Removal of indigestible material, bacteria and bile pigments from the body

Emulsify To mix two liquids that would not normally mix

Faeces Material passed out of the rectum

Gastric gland The secretory unit of the stomach located in gastric pits; produces gastric juice

Gastric juice The digestive juice secreted by the glands of the stomach

Incisor The narrow-edged tooth at the front of the mouth; used for cutting

Ingestion The intake of food, liquids or drugs into the mouth

Intestinal juice The digestive juice secreted by the glands of the small intestine

Lacteal A lymph capillary in the small intestine; it absorbs fat from digested food

Large intestine The part of the intestine between the small intestine and the anus; it is made up of the caecum, colon and rectum

Longitudinal muscle Smooth muscle with fibres arranged lengthwise along an organ

Mastication The process of chewing; to grind or crush food with the teeth

Mechanical digestion The mechanical breakdown of food into small particles

Microvilli Microscopic projections from the membranes of cells lining the small intestine; they increase the surface area for absorption; singular: microvillus

Molar A grinding tooth at the back of the mouth

Mucosa A mucous membrane; in particular, the mucous membrane that forms the internal lining of the alimentary canal

Oesophagus The tube that carries food from the throat to the stomach

Pancreatic amylase An enzyme in pancreatic juice that breaks down starch

Pancreatic juice The liquid secreted by the pancreas

Pancreatic lipase An enzyme in pancreatic juice that breaks down fats

Peristalsis Waves of muscular contraction that push food along the alimentary canal

9780170449090

Pharynx The throat; the pharynx joins the mouth cavity to the oesophagus and larynx

Premolars The teeth between the canine and molars

Pyloric sphincter A ring of smooth muscle between the stomach and the duodenum

Ribonuclease An enzyme in pancreatic juice that digests RNA

Saliva A fluid secreted into the mouth by salivary glands to begin digestion of food

Salivary gland Gland in the mouth that secretes saliva

Segmentation A process occurring in the small intestine which uses the contraction of circular muscles to push the chyme into segments, mixing it with digestive juices

Small intestine The longest part of the alimentary canal; receives material from the stomach

Stomach A muscular organ that receives food from the oesophagus, and mixes it with acid and enzymes to form chyme

Trypsin An enzyme in pancreatic juice that breaks down protein; also known as pancreatic amylase

Villi Projections from the internal lining of the small intestine; also, projections of the chorion that grow into the lining of the uterus; singular: villus

9780170449090

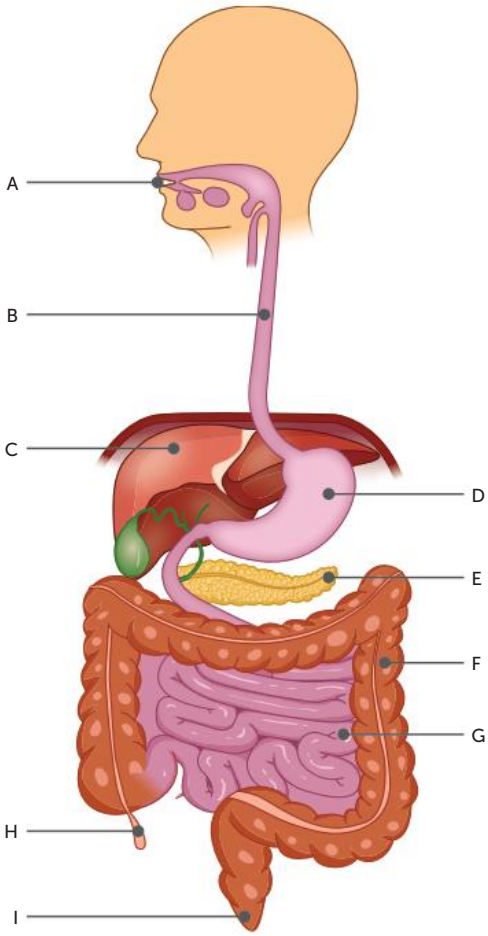
CHAPTER 6 REVIEW QUESTIONS

Recall

1 Draw up a table with three columns. In the first column, list the parts of the alimentary canal that are discussed in this chapter. In the second column, describe the role of each part in digestion and absorption. In the third column, explain how the structure of the part is suited to its functions. Remember to put an appropriate heading on each of your columns.

2 What are the basic activities that the digestive system must carry out?

3 Label the parts of the alimentary canal on the diagram below.



4 Name the enzyme(s) that break down:
- **a** proteins
- **b** complex carbohydrates
- **c** lipids.

Explain

5 Explain the digestive function of each of the following:
- **a** gastric juice
- **b** bile
- **c** pancreatic juice
- **d** intestinal juice.

6 Explain the difference between excretion and elimination.

9780170449090

7 To be effective, any surface where materials are taken into or passed out of the body must have a very large surface area. Explain how a large surface area is achieved in the part of the digestive system where nutrients are absorbed.

Apply

8 What effect would each of the following have on the speed with which food moves through the alimentary canal?
- **a** Consuming a very large meal
- **b** Eating a meal that is very high in fat
- **c** Consuming alcohol

9 **a** Describe the cause of constipation.
- **b** What precautions can you take to prevent constipation?

10 Why are people who suffer from coeliac disease likely to become malnourished?

Extend

11 Absorption of nutrients depends on concentration differences so that substances diffuse across the absorbing surface. Explain how the concentration difference is maintained in the parts of the alimentary canal where absorption occurs.

12 Explain why some food substances (such as starch) have to be digested but others (e.g. salt) do not.

13 The most common treatment for bowel cancer is to surgically remove the part of the bowel containing the cancer. Suggest what effects removal of part of the large intestine may have on a person's normal functions.

14 Use a flow chart to outline the process of digestion of a piece of ham, which is a protein.

9780170449090

7 THE EXCRETORY SYSTEM REMOVES WASTE PRODUCTS

UNIT 1 CONTENT

SCIENCE INQUIRY SKILLS

» conduct investigations, including monitoring body functions; use microscopy techniques; and perform real or virtual dissection, safely, competently and methodically for the collection of valid and reliable data

SCIENCE AS A HUMAN ENDEAVOUR

» lifestyle choices, including being active or sedentary, the use of drugs and type of diet, can compromise body functioning in the short term and may have long-term consequences

» treatment of conditions due to system or organ dysfunction has changed through improvements in early diagnosis and appropriate use of drugs, physical therapy, radiation therapy, and removal and/or replacement of affected parts

SCIENCE UNDERSTANDING

Excretory system

» the excretory system regulates the chemical composition of body fluids by removing metabolic wastes and retaining the proper amounts of water, salts, and nutrients; components of this system include the kidneys, liver, lungs, and skin functioning at the organ level

» deamination of amino acids in the liver produces urea, which then is transported to the kidneys for removal

» the nephrons in the kidney facilitate three basic processes: filtration, reabsorption and secretion during urine formation to maintain the composition of body fluids (hormone control is not required)

Source: School Curriculum and Standards Authority, Government of Western Australia

9780170449090

Chemical processes in the body produce more than just the desired product. These other chemicals are called by-products. Some by-products can be used by the body, while others are wastes. Most of the wastes are toxic and would be harmful to one's health if allowed to accumulate. Every cell produces waste products, so removing the wastes before they reach harmful concentrations is extremely important. Removal of the wastes of metabolism from the body is called **excretion**.

7.1 THE ORGANS THAT PROCESS AND REMOVE WASTES

Several organs in the body are involved either in the processing of wastes or in the excretion of those wastes.

- The **lungs** are involved in the excretion of the carbon dioxide that is produced by all body cells during cellular respiration. The removal of carbon dioxide by the lungs was discussed in Chapter 4.
- The **liver** has an important role in processing many substances so that they can be excreted.
- **Sweat glands** in the skin secrete sweat, which is largely water, for cooling. Sweat contains by-products of metabolism such as salts, urea and lactic acid.
- The alimentary canal passes out bile pigments, which enter the small intestine with the bile. These pigments are the breakdown products of haemoglobin from red blood cells.
- The **kidneys** are the principal excretory organs. They are responsible for maintaining the constant concentration of materials in the body fluids. The most toxic wastes removed by the kidneys are the nitrogenous wastes urea, uric acid and creatinine. Urea is produced in the liver from the breakdown of amino acids, which come from protein metabolism.

FIGURE 7.1 a The lungs, **b** liver, and **c** kidneys are involved in the processing and excretion of wastes

Key concept

The kidneys, liver, lungs, sweat glands and alimentary canal are all involved in processing and excreting wastes.

Questions 7.1

RECALL KNOWLEDGE

1 List the organs involved in excretion.
2 Describe how the alimentary canal is involved in excretion.
3 Summarise how amino acids are broken down and removed from the body.

APPLY KNOWLEDGE

4 Some people are living donors. This means that they are alive when they donate an organ such as a kidney to someone else. Explain how they are able to do this and still remain healthy.
5 People who consume excess alcohol have a higher risk of liver problems. Suggest why this is so.

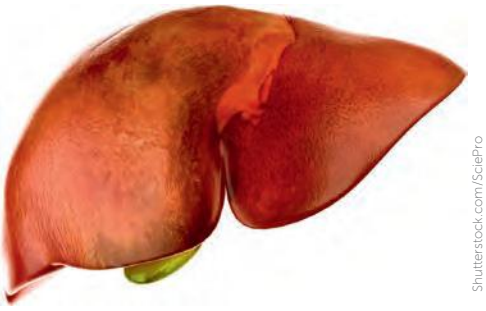
7.2 THE LIVER AND SKIN

The liver and the skin are the two heaviest organs in the body. They both play important roles in processing and excreting waste materials.

a

Shutterstock.com/sirtravelalot

b



Shutterstock.com/SciePro

FIGURE 7.2 a The skin; **b** A liver. On average, in a male adult, the skin weighs 4.5 kg and the liver 1.6 kg

The liver

The liver is located in the upper abdominal cavity. It is a very large organ with a host of different functions, one of which is the preparation of materials for excretion.

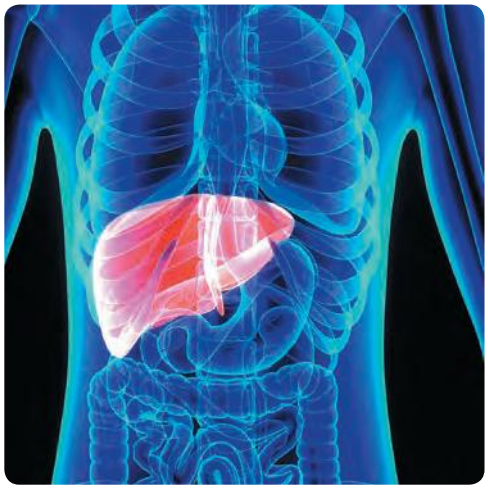
Proteins are primarily 'body builders'; that is, they make up the structural materials of cells. As long as the body has a sufficient supply of carbohydrates and fats, or has a supply of stored fat, then little protein is used in energy-releasing reactions. Excess protein from the diet cannot be stored in the cells, and so processes are required to remove it from the body.

Some protein is broken down in the body all the time, although most of the breakdown is incomplete. Worn-out cells, such as red blood cells, are a source of protein, and are broken down into the constituent amino acids. Most of these amino acids are then used to make new proteins. However, a very small amount is lost from the body via the urine, skin, hair and fingernails.

The proteins, which have been built up from amino acids, become the primary constituents of cell structures, enzymes, antibodies and many glandular secretions. However, if other energy sources have been used up, the body is able to metabolise large amounts of proteins, breaking them down to produce energy.

To make use of proteins in this way, the amino group (NH_2) must first be removed from the amino acids. This process, called **deamination**, occurs in the liver with the aid of enzymes. Once the amino group has been removed, it is converted by the liver cells to ammonia (NH_3) and then finally to urea. The urea is eliminated from the body in **urine**. The remaining part of the amino acid, which is primarily made up of carbon and hydrogen, is converted into a carbohydrate. This carbohydrate can be readily broken down by the cells to release energy, carbon dioxide and water.

3D image of the liver



Shutterstock.com/sciencepics

FIGURE 7.3 Position of the liver in the body

Deamination can be summarised as an equation:

$$\text{amino acid + oxygen} \xrightarrow{\text{enzymes}} \text{carbohydrate + ammonia}$$

Ammonia is extremely soluble in water and is highly toxic to cells. One-thousandth of a milligram of ammonia in a litre of blood is sufficient to kill a person. The cells of the liver rapidly convert ammonia to the less toxic molecule urea. Moderate amounts of urea are harmless to the body. It is easily excreted by the kidneys and is eliminated from the body in the urine. Small amounts of urea are also lost in sweat from the sweat glands. The process can be expressed as:

$$\text{energy + carbon dioxide + ammonia} \longrightarrow \text{urea + water}$$

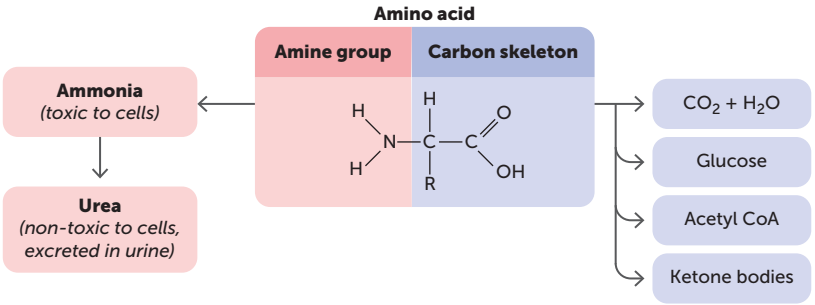


FIGURE 7.4 The breakdown of amino acids

The liver also:

- detoxifies alcohol and many other drugs such as antibiotics
- deactivates many hormones and converts them into a form that can be excreted by the kidneys
- breaks down haemoglobin from dead red blood cells to produce bile pigments, which are then passed out of the body with the faeces.

Key concept

The liver plays an important role in processing chemicals into a safer form. For example, it converts ammonia produced from proteins into the safer form of urea.

The skin

The main functions of the skin are to provide a protective covering over the surface of the body and to regulate body temperature. However, skin also has an important role in excretion.

Even when there is no visible perspiration on the skin, the sweat glands secrete about 500 mL of water per day. Dissolved in the water are sodium chloride, lactic acid and urea. These substances are being excreted from the body. Some drugs, such as salicylic acid, are also excreted by the skin.

Sweat glands are located in the lower layers of the skin. A duct carries the sweat to a hair follicle or to the skin surface where it opens at a pore. Cells surrounding the glands are able to contract and squeeze the sweat to the skin surface.

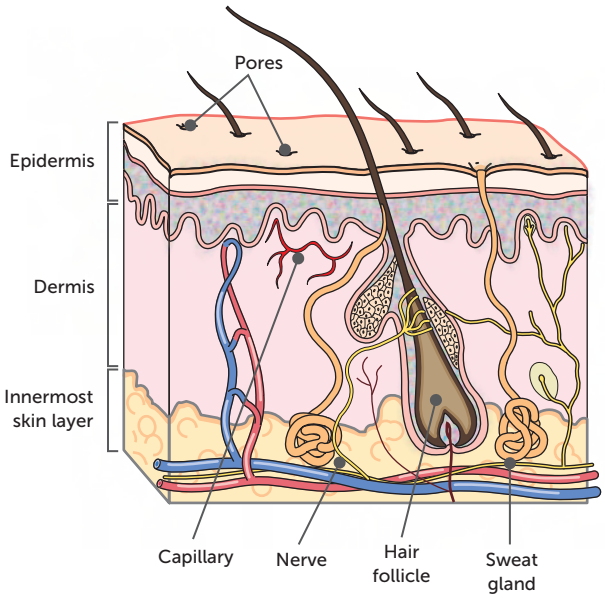


FIGURE 7.5 The structure of skin, including sweat glands

Questions 7.2

RECALL KNOWLEDGE

1 Define 'deamination'.
2 Name the functions of the skin in excretion.
3 List three substances that the liver processes for excretion.
4 Describe what happens to proteins that are broken down.

APPLY KNOWLEDGE

5 Patients with liver disease have higher than normal levels of ammonia in their blood. Explain why this occurs.

7.3 THE KIDNEYS

The **kidneys** are a pair of reddish-brown organs located in the abdomen. Each kidney is approximately 11 cm long. Their position and relative size can be seen in Figure 7.6. The kidneys, the bladder and their associated ducts are often referred to as the **urinary system**.

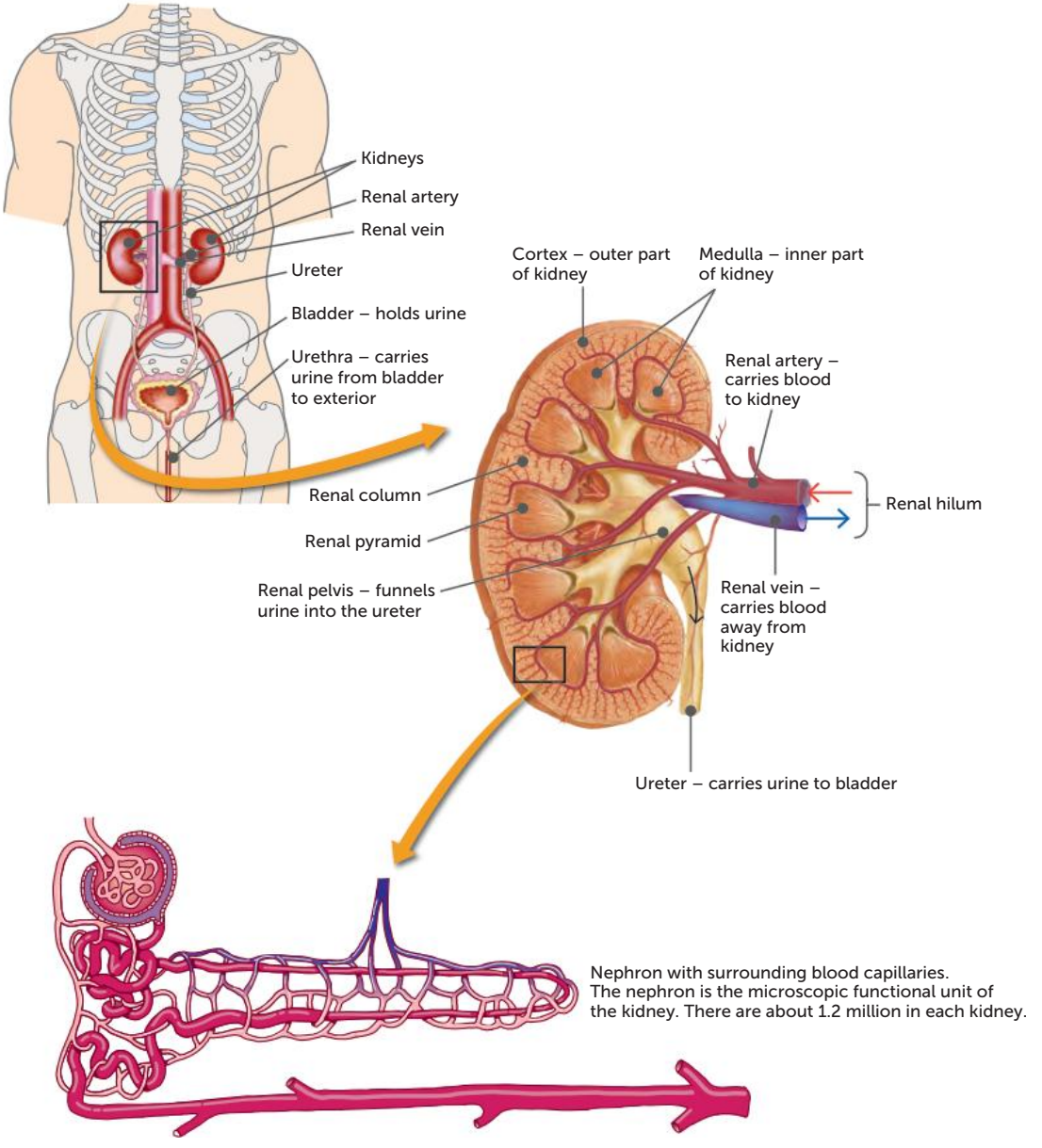


FIGURE 7.6 Structure and functions of the organs of the urinary system

Activity 7.1
Examining the structure of the kidneys: a dissection

9780170449090

The kidney is enclosed by the **renal capsule**. Under this is the outer **renal cortex**, the inner **renal medulla**, and then the **renal pelvis** sits in the concave side of the kidney. The **renal hilum** lies on the concave surface of the kidney, and is where the vessels enter and leave. The medulla consists of a number of **renal pyramids**, which are separated by **renal columns**, where the blood vessels lie.

The structure of nephrons

When examined under a microscope, the kidney is seen to be composed of a large number of structures called **nephrons** and **collecting ducts**. The nephron is the functional unit of the kidney, as it is where the urine is formed. There are about 1.2 million nephrons in each human kidney, and each is surrounded by a complex network of blood capillaries.

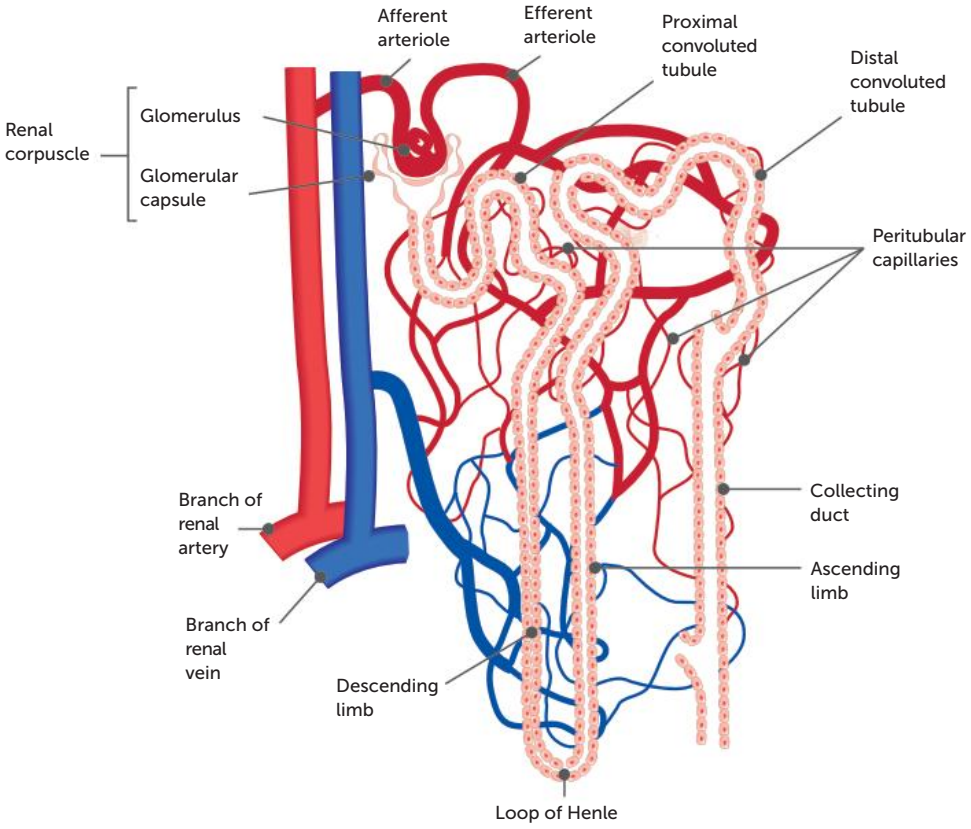


FIGURE 7.7 A nephron, showing arrangement of the blood vessels

Learn about the kidney

Key concept

The nephron is the microscopic, functional unit of the kidney.

Each nephron consists of a **renal corpuscle** and a **renal tubule**. The renal corpuscle consists of the glomerulus and the glomerular capsule. The **glomerular capsule** (formerly known as the Bowman's capsule) is the expanded end of the nephron. It looks like a double-walled cup that surrounds, and almost completely encloses, a knot of arterial capillaries called the **glomerulus**.

Leading away from the glomerular capsule is the renal tubule, a tube about 5 cm long. It begins with a winding, or convoluted, section called the **proximal convoluted tubule**. Beyond this, each tubule has a straight portion before it forms a loop, the **loop of Henle**. The loop of Henle is like a hairpin bend, with a straight section leading into the bend and another straight section leading away from the bend. The tubule then becomes convoluted and highly coiled again. This second coiled section is known as the **distal convoluted tubule**. The distal convoluted tubules of several nephrons join into a **collecting duct** that opens into a chamber in the kidney called the renal pelvis. The renal pelvis is shaped like a funnel and channels fluid from the collecting ducts into the **ureter**.

9780170449090

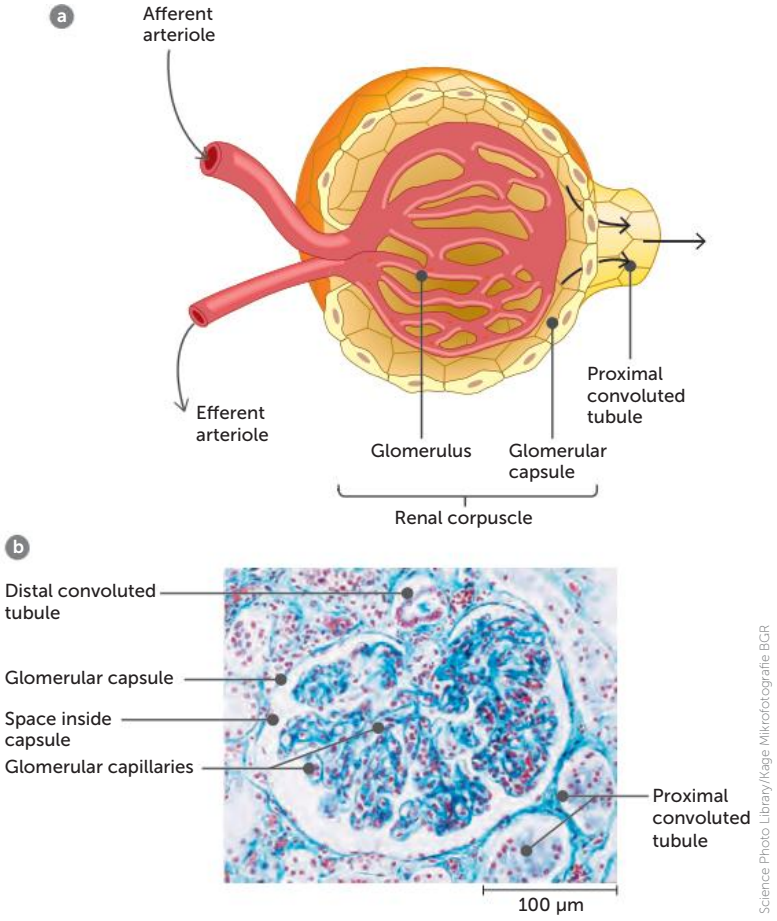


FIGURE 7.8 a The renal corpuscle; **b** Photomicrograph showing a renal corpuscle

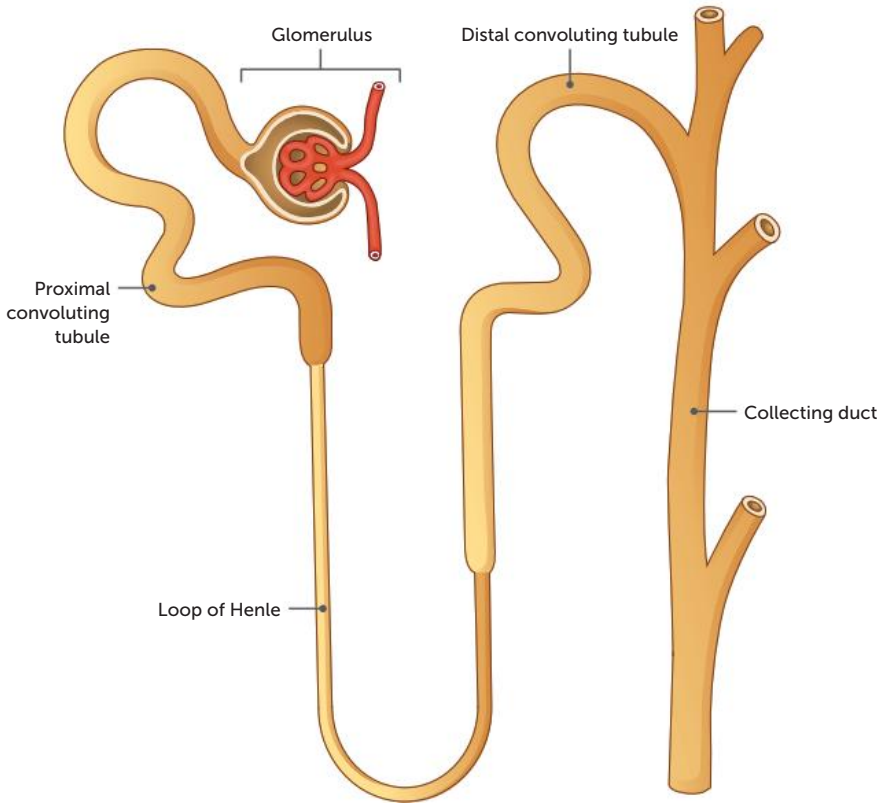


FIGURE 7.9 The renal tubules

Activity 7.2
Looking at nephrons

The nephrons of the kidney are responsible for removing wastes from the blood and regulating blood composition. To be able to do this, they are well supplied with blood vessels. Blood enters the kidney through the **renal arteries**. These arteries are quite large, so together the two kidneys receive about a quarter of the blood from the heart. Approximately 1.2 L of blood pass through the two kidneys every minute.

Shortly after entering the kidney, the renal artery divides into small arteries and arterioles. Each renal corpuscle is supplied by an arteriole, the **afferent arteriole**, which then forms a knot of capillaries called the glomerulus. This knot of capillaries is located within the glomerular capsule. The capillaries eventually unite to form another arteriole, the **efferent arteriole**, which passes out of the renal corpuscle.

After leaving the renal corpuscle, the efferent arteriole breaks up into a second capillary network. These capillaries surround the proximal and distal convoluted tubules of the nephron, the ascending and descending limbs of the loop of Henle, and the collecting duct. They are known as **peritubular capillaries** (Figure 7.7). Venous blood drains away from this network of capillaries and eventually leaves the kidney in the **renal vein**.

Key concept

Blood enters the nephron through the afferent arteriole. It is filtered in the glomerulus, a network of capillaries, and then exits via the efferent arteriole.

The production of urine

The formation of urine by the nephrons of the kidneys involves three major processes: **glomerular filtration**, **selective reabsorption** and **secretion by the tubules**.

Glomerular filtration

The first step in the production of urine is glomerular filtration. This process takes place in the renal corpuscle when fluid is forced out of the blood and is collected by the glomerular capsule. Fluid is normally forced out of the capillaries into the tissue in all parts of the body due to differences in pressure between the capillaries and tissue. In the glomerulus the process is enhanced by the high pressure of blood. The afferent arteriole leading into the glomerulus has a wider diameter than the efferent arteriole leaving it. This narrowing of the efferent arteriole increases resistance to the flow of blood and produces a higher pressure in the glomerulus.

The blood in the capillaries in the glomerulus is separated from the cavity of the capsule by only two single layers of thin, flat cells. One layer makes up the capillary wall and the other the wall of the capsule. Therefore, when blood enters the glomerulus, the high pressure forces water and dissolved blood components through the differentially permeable cell membranes and into the capsule. The resultant fluid is termed the **filtrate**.

In a healthy person, the filtrate consists of all the materials present in the blood except red and white blood cells and plasma proteins. These are too large to pass through the differentially permeable membranes of the cells making up the walls of the glomerulus and capsule. Therefore, the filtrate consists of water, salts, amino acids, fatty acids, glucose, urea, uric acid, creatinine, hormones, toxins and various ions.

As blood flows through the capillaries of the glomerulus, 20% of the plasma is filtered through the capillary walls into the glomerular capsule. Complete filtration of all the plasma cannot take place as the blood in the capillaries is continually being pushed on by the blood behind it. With about 1.2 million nephrons in each kidney, the amount of plasma filtered every minute is still quite high. In normal adults, the total filtrate produced by all the renal corpuscles of both

kidneys is about 125 mL of filtrate per minute. This amounts to about 180 L in a day! Although this large amount is filtered, only about 1% actually leaves the body as urine. Most is reabsorbed back into the blood.

Reabsorption

Many of the components of the plasma that are filtered from the capillaries of the glomerulus are of use to the body, and their excretion would be undesirable. Therefore, some **selective reabsorption** of the filtrate must take place, returning them to the blood in the peritubular capillaries. These processes are carried out by the cells that line the renal tubule. Materials that are reabsorbed include water, glucose and amino acids. Ions such as sodium, potassium, calcium, chloride and bicarbonate are also reabsorbed. Some wastes, such as urea, are partially reabsorbed as well.

Like the body's other exchange surfaces, a large surface area is required. This large surface area for effective reabsorption of materials is achieved by the long length of the kidney tubule – two sets of convolutions and the long loop of Henle – and by the huge number of nephrons in each kidney.

Reabsorption of much of the water in the filtrate can be regulated. Depending on the body's water requirements, the permeability of the plasma membranes of the cells making up parts of the tubules can be changed. Therefore, more or less water can be reabsorbed depending on the body's requirements. This is an active process, under hormonal control, and is often referred to as **facultative reabsorption**. This is discussed further in Unit 3.

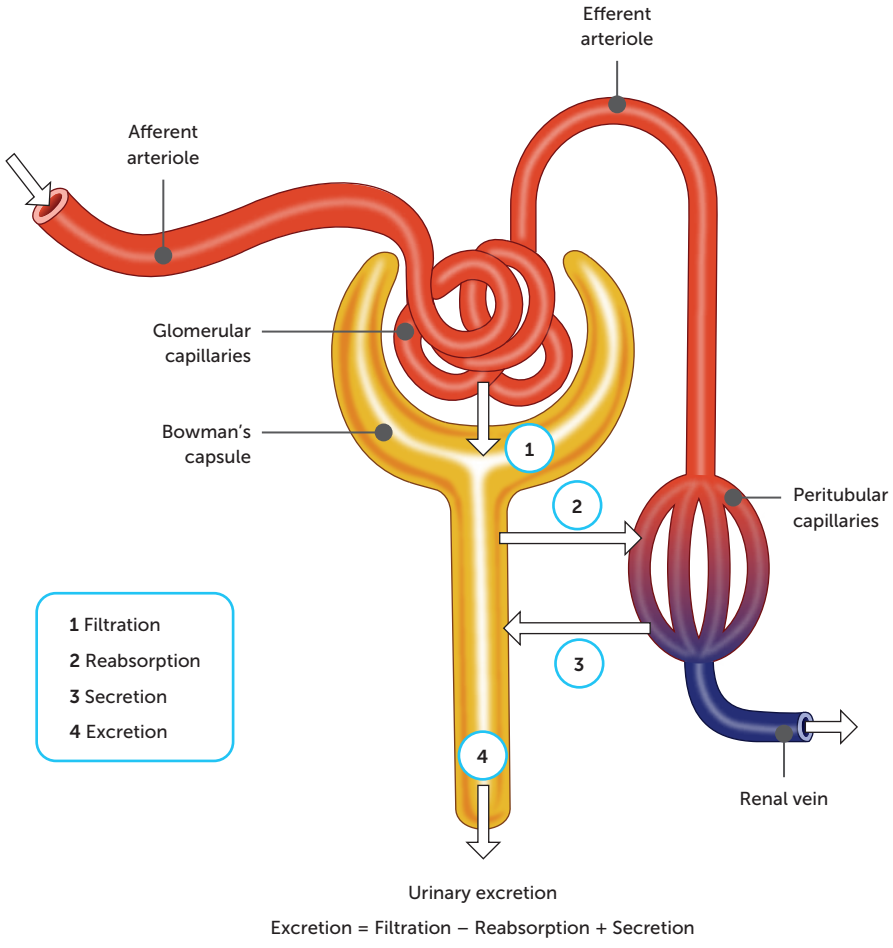


FIGURE 7.10 The process of filtration, reabsorption and secretion

Tubular secretion

The third process involved in the formation of urine is **tubular secretion**. Whereas selective reabsorption *removes* substances from the filtrate into the blood, tubular secretion *adds* materials to the filtrate from the blood, as Figure 7.10 shows. Materials secreted in this way include potassium and hydrogen ions, creatinine, and drugs such as penicillin.

Tubular secretion can be either active or passive, and has two main effects.

- It maintains the blood pH. The body needs to maintain the blood within its normal pH range of 7.4–7.5. Our diets usually contain many acid-producing foods that tend to lower pH; therefore, the kidneys must remove the excess hydrogen and ammonium ions.
- It maintains the urine pH. The presence of hydrogen and ammonium ions in the urine makes the urine slightly acidic, with a normal pH of 6.

Key concept

The processes of glomerular filtration, selective reabsorption and tubular secretion control the composition of urine and, therefore, the blood.

Water and other substances not reabsorbed drain from the collecting ducts into the renal pelvis. From the pelvis, the urine, as it is now called, drains into the ureters and is pushed by waves of muscle contraction to the urinary bladder where it is stored. The two ureters, one from each kidney, are essentially extensions of the pelvis of the kidneys. They extend 25–30 cm to the urinary bladder. The bladder is a hollow muscular organ from which the urethra exits. The urethra carries urine from the bladder to the exterior of the body.

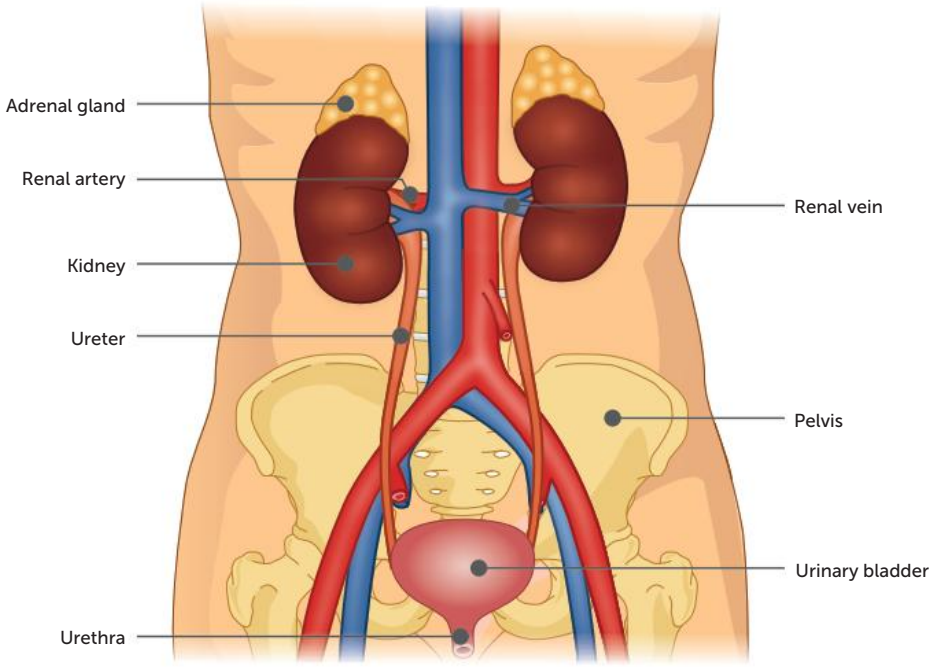


FIGURE 7.11 Structures of the urinary system

The formation of urine is maximised by the structure of the kidney, particularly the nephron, as follows:

- The glomerular capsule surrounds the glomerulus to collect the fluid filtered out of the blood capillaries.
- There are only two cells for the filtrate to pass through from the blood, one cell from the capillary wall and the other from the capsule wall.

- A large volume of blood passes through each kidney. The continual flow maintains the concentration gradient.
- The efferent arteriole leading out of the glomerulus has a smaller diameter than the afferent arteriole leading in. This raises the blood pressure so that more fluid is filtered out of the blood.
- Each tubule has a large surface area for reabsorption and secretion due to each tubule having two sets of convolutions and a long loop.
- Each kidney has over a million nephrons, so the total surface area available for reabsorption and secretion is extremely large.

TABLE 7.1 Summary of the functioning of the kidney

REGION OF NEPHRON	ACTIVITIES TAKING PLACE
Renal corpuscle	Filtration of blood from capillaries of glomerulus
	Formation of filtrate in the glomerular capsule
Proximal convoluted tubule and loop of Henle	Passive reabsorption of potassium, chloride and bicarbonate ions
	Active reabsorption of glucose and sodium
	Passive reabsorption of water by osmosis
Distal convoluted tubule	Active reabsorption of sodium ions
	Active reabsorption of water, depending on the body's water needs
	Secretion of hydrogen and potassium ions, creatinine and certain drugs such as penicillin
Collecting duct	Active reabsorption of water, depending on the body's water needs

7.1 The kidneys

The composition of urine

The body must excrete its waste products, such as urea, sulfates and phosphates, on a regular basis. These substances need to be in solution, and so the elimination of these wastes requires a certain amount of water loss. Regardless of the amount of water available in the body, or the amount taken in, about half a litre of water must be lost each day simply to remove wastes. When the water content of the body fluids is low, the urine that is excreted is very concentrated.

Table 7.2 compares the composition of the fluid filtered from the blood and the urine. It also shows the amount of each component that is reabsorbed during a 24-hour period. The values shown can vary markedly between individuals, and with diet, and so they should be regarded only as a guide.

TABLE 7.2 Composition of filtrate, reabsorbed substances and urine over a 24-hour period

CHEMICAL COMPONENT	FILTRATE	REABSORBED SUBSTANCES	URINE
Water	180 L	178–9 L	1–2 L
Sodium, chloride and other ions	1500 g	1485 g	15 g
Proteins	2 g	1.9 g	0.1 g
Glucose	180 g	180 g	0 g
Urea	53 g	28 g	25 g
Uric acid	8.5 g	7.5 g	1 g
Creatinine	1.6 g	0 g	1.6 g

Activity 7.3
Investigating kidney output

Uric acid is produced by the metabolism of substances called purines. Purines may come from the breakdown of nucleic acids, such as DNA, when cells die. Purines also occur naturally in many foods. **Creatinine** is produced in muscle from the breakdown of creatine phosphate, an energy-rich molecule. Thus, under normal circumstances:

- about 99% of the water that enters the nephron is reabsorbed
- the urine does not normally contain significant amounts of protein
- the urine does not normally contain any glucose
- the main materials making up the urine, besides water, are urea, ions, uric acid and creatinine.

Generally, a healthy adult produces about 1.5 L of urine a day, but this varies tremendously depending on diet, environment and other factors. Table 7.3 indicates the components that the urine of a healthy adult may contain. Again, this is highly variable and should be used only as a guide. The amber colour of urine is due to the presence of some bile pigments.

Activity 7.4
Investigating urine concentration

Activity 7.5
Modelling kidney function

TABLE 7.3 Composition of urine

COMPONENT	%
Water	96.0
Urea	2.0
Various ions	1.5
Other	0.5

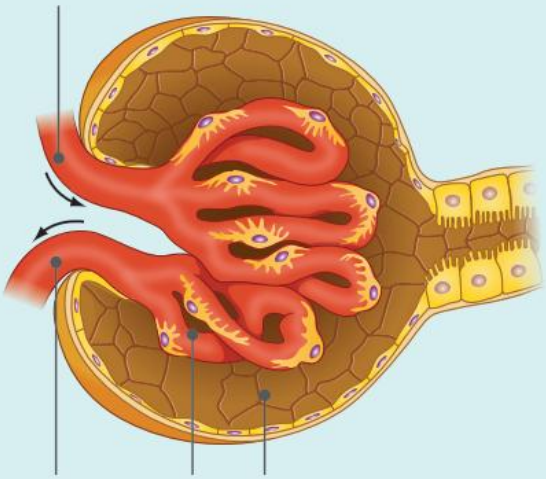
Key concept

Normal urine is a solution of water with dissolved wastes, such as urea and creatinine, and ions such as sodium, chloride and potassium, as well as low levels of other solutes.

Questions 7.3

RECALL KNOWLEDGE

1 Label the diagram of a renal corpuscle.



2 List the components of the filtrate that are reabsorbed.

3 State where each of the following processes occurs:
   a filtration
   b secretion
   c reabsorption
   d storage of urine
   e drainage of urine from the nephrons.

4 Draw a diagram of a cross-section of the kidney and label the:
   a renal artery and renal vein
   b renal medulla
   c renal cortex
   d renal pyramids
   e renal capsule
   f renal columns
   g renal pelvis.

→

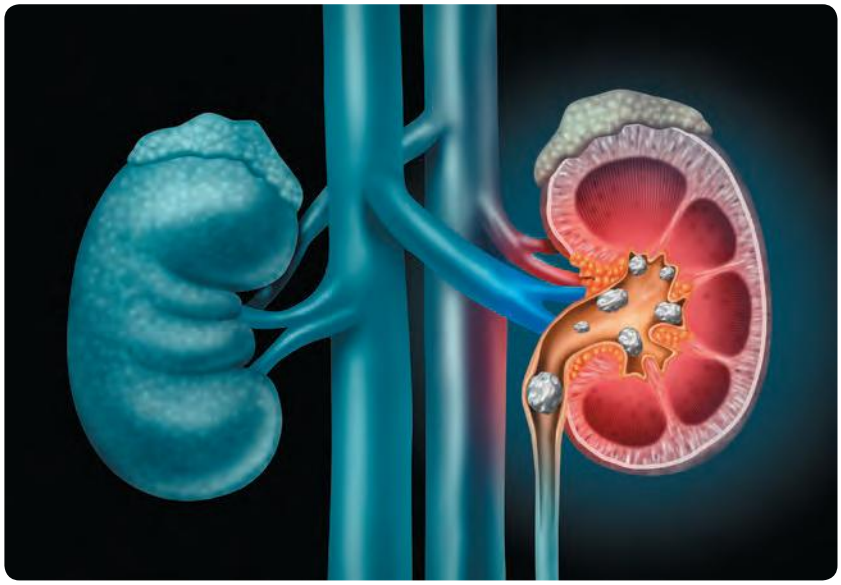
APPLY KNOWLEDGE

5 Explain the importance of reabsorbing glucose.
6 Explain why the active reabsorption of water is vital in maintaining the correct fluid levels in the body.
7 Describe how the kidney is able to have a large surface area for the processes involved in excretion.
8 Compare and contrast the process of glomerular filtration with the movement of fluid from capillaries in other body tissues.

7.4 EFFECTS OF LIFESTYLE ON EXCRETION

Kidney stones

Kidney stones are formed from solid crystals that build up inside the kidneys. They usually form when urine becomes too concentrated. Some doctors believe that kidney stones are caused by insufficient fluids in the diet. If the crystals are small enough, they may pass down the ureter and out of the body through the urethra without being noticed. Crystals may combine to form stones. Large stones may get stuck in the ureter, bladder or urethra, causing intense pain. Smaller kidney stones, while painful, may pass with the aid of fluids, pain relief and muscle relaxants. Larger stones, however, may need to be broken up with sound waves or physically removed during surgery.



Shutterstock.com/Lightspring

FIGURE 7.12 A kidney with kidney stones

Kidney failure

One in three adult Australians is at risk of developing kidney disease. A person can lose up to 90% of kidney function without realising it. But by that stage it is almost impossible to prevent serious problems occurring.

Most kidney diseases affect the glomeruli, reducing their ability to filter the blood. Protein and sometimes red blood cells may leave the blood at the glomerulus and will then be present in the urine. If excessive proteins are lost in the urine, blood protein levels fall and fluid accumulates in the tissues, causing swelling of the hands, feet, face or other areas.

There are a number of lifestyle measures that you can take to maintain healthy kidneys.

- Regulate your diet to maintain a healthy weight. Being overweight can lead to the development of diabetes or high blood pressure, both of which are major risk factors for kidney disease.
- Do not smoke. Compared with non-smokers, people who smoke are three times more likely to have impaired kidney function.

9780170449090

- Drink water instead of drinks containing sugar.
- Drink alcohol in moderation – one standard drink per day for women and two per day for men.
- Do not use performance-enhancing drugs, as they may upset the water balance in the body. In particular, anabolic steroids can cause scarring inside the kidneys and eventual kidney failure.

When the kidneys lose their ability to excrete waste and to control the level of fluid in the body, this is known as **kidney failure**. Kidney failure may happen suddenly, but is more likely to develop over a period of years. Factors such as diabetes, high blood pressure or kidney diseases slowly destroy the nephrons in the kidneys. Eventually, the only way to maintain life is by dialysis or a kidney transplant.

Dialysis

Dialysis is a method of removing wastes from the blood when kidney failure occurs. There are two types of dialysis: peritoneal dialysis and haemodialysis.

The **peritoneum** is a membrane that lines the inside of the abdominal cavity and covers abdominal organs such as the stomach, liver and intestines. It has a very rich blood supply. **Peritoneal dialysis** occurs inside the body using the peritoneum as a membrane across which waste can be removed. A tube, called a catheter, is placed through the wall of the abdomen. For an adult, 2–3 L of fluid are passed through the catheter into the abdominal cavity. The fluid contains glucose and other substances at concentrations similar to those found in the blood. However, there are no wastes in the fluid. This means that, because of the concentration difference, wastes will diffuse out of the blood into the fluid in the abdominal cavity. Useful substances stay in the blood because there is no concentration difference between the blood and the fluid. After a time, the fluid that was placed in the abdominal cavity is drained out through the catheter, along with any wastes and extra water that have diffused from the blood. Peritoneal dialysis is usually done each day.

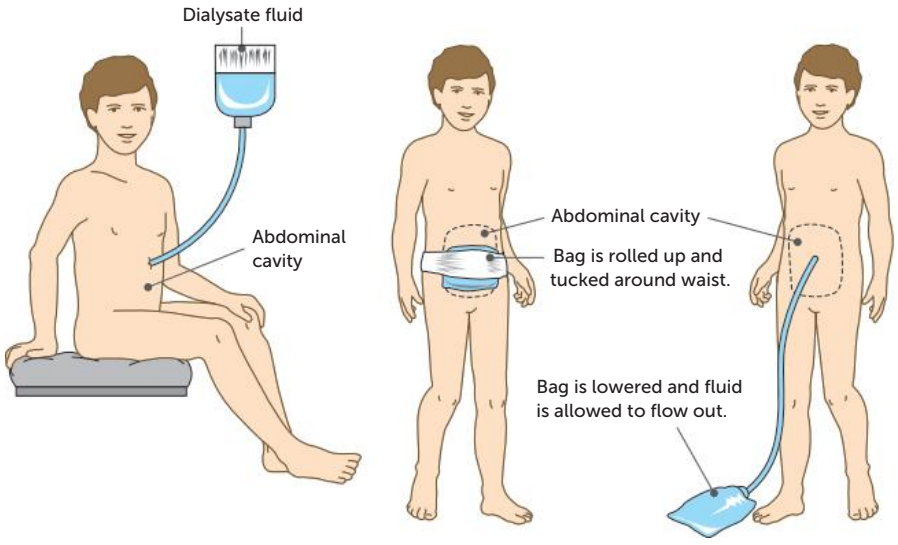


FIGURE 7.13 The components of peritoneal dialysis

Dialysis
This website shows how dialysis works.

Haemodialysis involves passing the blood through an artificial kidney or dialysis machine. The blood passes through thousands of fine tubes, made of a differentially permeable membrane, and immersed in a bath of fluid. The concentrations of substances in the fluid are similar to those in the blood, except that the fluid has no waste. Because of the concentration differences, wastes diffuse from the blood into the fluid. Patients spend about 4–5 hours attached to the machine and dialysis is normally done three times per week.

9780170449090

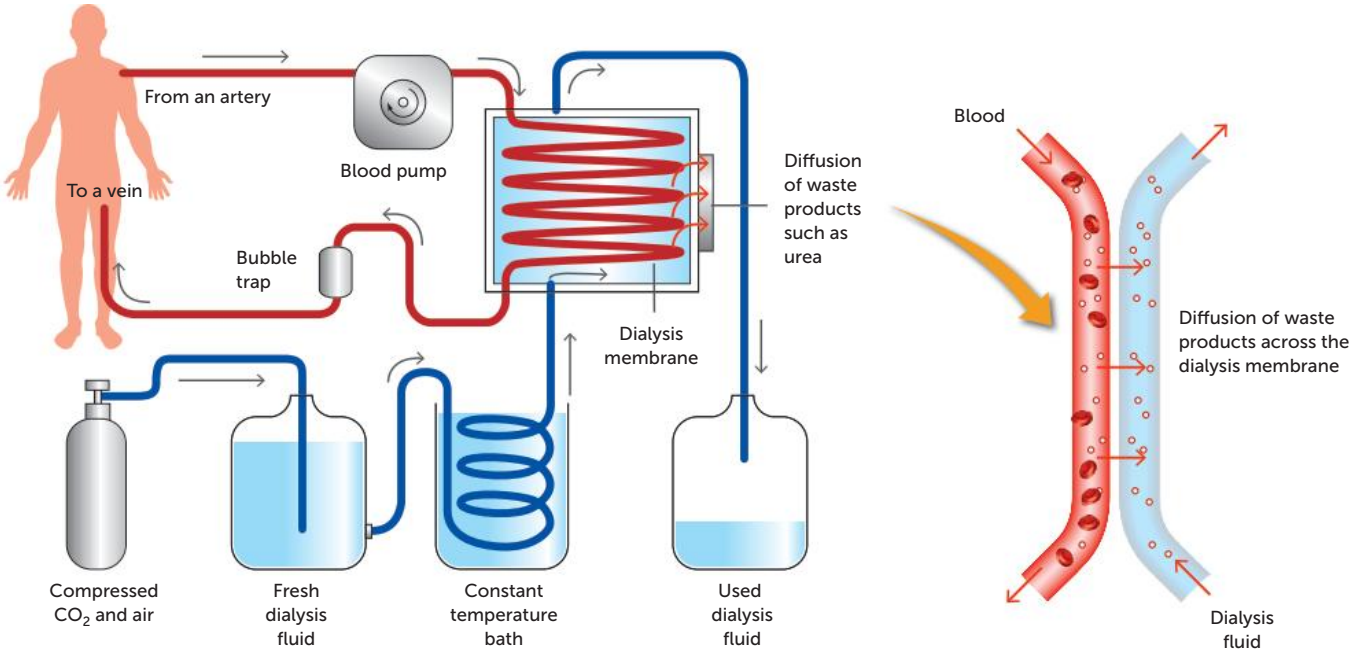


FIGURE 7.14 The components of haemodialysis

Liver disease

When the liver is not able to function effectively, it is unable to process toxins ready for elimination. Liver disease can be caused by infection, autoimmune problems, genetic disorders, cancer, and lifestyle factors such as excessive alcohol consumption and a fatty diet.

People who suffer from liver disease show symptoms due to a build-up of the toxins that would normally be eliminated. These include a yellow tinge to the skin (called jaundice), abdominal pain and swelling, swelling in the legs and feet, nausea or vomiting, fatigue, dark urine, and faeces that are pale or dark coloured.

Key concept

Lifestyle choices such as diet, alcohol consumption, water intake and drugs can lead to damage to the kidneys and liver. This can have an impact on the effectiveness of excretion.

Questions 7.4

RECALL KNOWLEDGE

1. List the types of dialysis.
2. List the causes of liver disease.
3. Describe kidney stones.

APPLY KNOWLEDGE

4. Explain how dialysis allows patients with kidney disease to live relatively normal lives.
5. Some patients with kidney stones require surgery to remove them. Explain what might happen if the kidney stones were not removed.

9780170449090

CHAPTER 7 ACTIVITIES

ACTIVITY 7.1 Examining the structure of the kidneys: a dissection

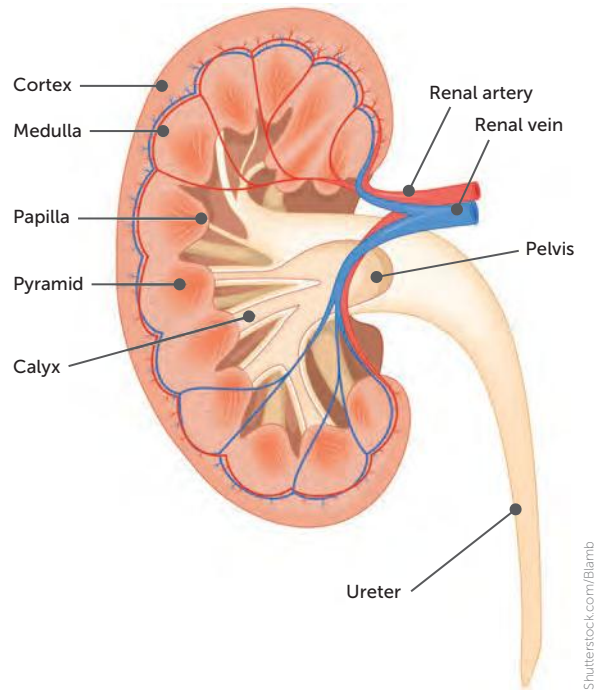
We can learn a lot by dissecting an organ and observing its structure. In this activity, you will dissect the kidney of a sheep so that you can gain a better understanding of the structure of human kidneys.

You will need

Kidney of a sheep; dissecting board; dissecting instruments; disposable gloves

What to do

1. Hold the kidney in your gloved hands. Feel the texture of the tissue.
2. Carefully remove any fat.
3. Identify the hilum of the kidney and any vessels that are present (renal artery, renal vein and ureter).
4. Carefully remove the renal capsule. Observe its thickness and appearance.
5. Use a scalpel to cut the kidney longitudinally in half, as shown below.



6. Identify the cortex, medulla, pyramids and pelvis.
7. Insert a probe into the base of a pyramid. Follow the calyx to the renal pelvis.
8. Dispose of the kidney as per your teacher's instructions.

Recording your observations

1. Describe the texture and external appearance of the kidney.
2. Draw a sketch, or take a photo, of the intact kidney. Label the hilum and any vessels that were present.
3. Describe the renal capsule.

4 Draw a sketch, or take a photo, of the longitudinal section of the kidney. Label any structures that you were able to identify.
5 Describe the pathway of the urine from the collecting duct of the nephrons to the ureter.

ACTIVITY 7.2 Looking at nephrons

In this activity, you will observe the microscopic structure of the kidney.

You will need

Microscope and microscope lamp; prepared slides of kidney tissue

What to do

When looking at cells on the prepared slides, remember that they have been stained to show up the structure of the cell and its contents.

1 Set up the light microscope (refer to Activity 2.1).
2 Place the slide of kidney tissue on the stage.
3 Use the lowest objective lens to focus on the slide.
4 Draw a diagram of what you can see. Label any structures that you can identify, such as the glomerular capsule and glomerulus.
5 Calculate the magnification and write this on your diagram.
6 Change the objective lens to a higher magnification.
7 Repeat steps 4 and 5 for this magnification.

ACTIVITY 7.3 Investigating kidney output

Table 7.2 (page 178) shows the amount of water and other substances filtered out of the blood of a healthy person in one day.

Questions

Use the information in the table, and your knowledge of how the kidney functions, to answer the following questions.

1 If 180 L of water are filtered out of the blood in 24 hours, why don't we produce 180 L of urine per day?
2 Why is there no protein in the fluid that is filtered out of the blood?
3 A large quantity of glucose is filtered out of the blood, but there is none in the urine. What happens to the glucose that is filtered out of the blood?
4 Of the substances listed in the table, which ones would be considered wastes? That is, for which of the substances does a high proportion of the filtered amount end up in the urine?
5 Suggest how the figures in the table might change if the person drank a large volume of water.
6 Table salt is sodium chloride. If a person ate very salty foods, what changes might be seen in the figures in the table?
7 Urea is formed when proteins are broken down in the liver. Suggest how the figures in the table might change if the person had a high-protein diet.

ACTIVITY 7.4 Investigating urine concentration

The higher the quantity of dissolved substances in a given volume of urine, the heavier it is. Therefore, a simple way of determining the concentration of urine is to weigh it. One litre of pure water has a mass of 1000 g; 1 L of urine has a mass of more than 1000 g because urine contains dissolved substances. Analysis of a person's urine by measuring the volume and recording the mass gave the results shown in the table on the next page. The results have

9780170449090

been converted to a standard volume of 1 L so that they can be directly compared. The higher the mass, the more concentrated the urine.

TIME	URINE MASS (g)
6.30 a.m.	1073
8.45 a.m.	1026
10.30 a.m.	1049
1.00 p.m.	1062
3.15 p.m.	1078
5.00 p.m.	1033
7.00 p.m.	1014
10.15 p.m.	1022

The results shown here were collected in summer and the person worked outdoors during the day.

Analysis of results

1 Using an appropriate format, draw a graph of these results.
2 For each of the measurements taken during the day, explain why the urine concentration has increased or decreased.

SOUTHERN Biological Developed exclusively by Southern Biological

ACTIVITY 7.5 Modelling kidney function

While the filtration that occurs within the kidney is far more controlled, this investigation provides a great illustration of how filtration occurs in the production of urine.

Aim

To investigate, by creating a model of the kidney, how the circulatory system and kidneys interact to create urine.
Time requirement: 50 minutes

You will need

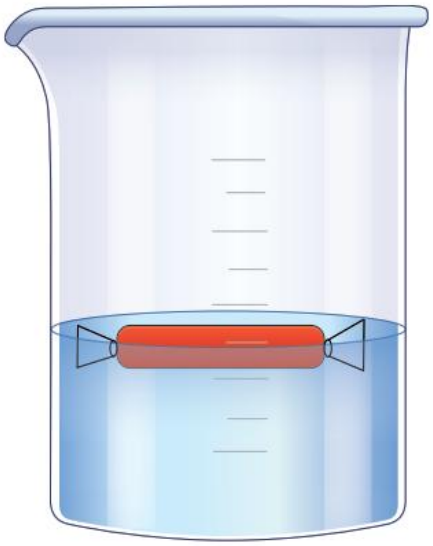
2 pipettes; 2 microscope slides; 2 coverslips; 250 mL glass beaker; 3 simulated salt test strips; 10 mL simulated kidney blood; 20 cm section of dialysis tubing (pre-soaked); 25 mL graduated cylinder; bulldog clip or similar; clock or timer; disposable gloves

Risks

WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
Staining from simulated kidney blood	Simulated kidney blood will stain skin and clothing. Avoid any direct contact with skin and clothing and wear appropriate personal protective equipment (PPE), such as gloves and lab coat.
Glass breakage	Inspect and discard any chipped or cracked glassware, no matter how minor the damage. Sweep up broken glass with brush and dustpan; do not use fingers.

What to do

1. Pour 100 mL of water into the beaker.
2. Place the tip of the salt test strip into the water for 3 seconds and gently swirl. To remove excess water, lightly tap the strip on the edge of the beaker or gently pat dry again on a paper towel. After 3 minutes, determine the results and record them in the data table.
3. Tie one end of the dialysis tubing into a knot. Ensure it is pulled tight, but do not allow the tubing to tear.
4. Measure 10 mL of simulated blood using a graduated cylinder and carefully pipette into the dialysis tubing. Once filled, twist the tubing to close the end and seal using a bulldog clip.
5. To ensure there is no simulated blood on the surface of the prepared dialysis tubing model, rinse it with tap water.
6. Place the dialysis tubing model into the beaker and add additional water until the filled portion of the dialysis tubing is fully submerged. Allow to rest in place for 30 minutes.



7. Add one drop of simulated blood onto a microscope slide using a pipette. Place a coverslip on top and observe under the microscope. Draw a sketch of what you observe in your notebook.
8. After 30 minutes, test the salt content of the filtrate using a new test strip. Place the strip in for 3 seconds and swirl gently. Remove excess water and wait 3 minutes to determine the results. Record the salt content results and any colour changes in the data table.
9. Add one drop of filtrate onto a new microscope slide using a pipette and place a coverslip on top. Observe the slide under the microscope and sketch what you see in your notebook.

9780170449090

Studying your results

Copy and complete the table below.

TEST	HYPOTHESIS	OBSERVATION
Initial water colour	N/A	
Filtrate colour (after 30 minutes)		
Initial colour dialysis tube contents	N/A	
Colour dialysis tube contents (after 30 minutes)		
Salt in plain water		
Salt in filtrate (after 30 minutes)		

Discussion

1. Which portion of the nephron were you modelling in this procedure? Explain.
2. Why was it necessary to test the water for salt before you added the dialysis tubing?
3. Imagine you left the tubing model in the cup for two days; how would the salt content in the filtrate change?
4. Compare the dialysis tubing model and kidneys. Examine how the model you created functioned like a kidney and the ways in which it did not.
5. What did the dialysis tubing retain? What was able to pass through the membrane?
6. Explain the difference between the solution within the dialysis tubing and surrounding water that allowed substances to travel through the membrane.

Taking it further

1. Kidney disease can be described as a systemic disease despite directly affecting the kidneys. In what ways can the whole body be affected by kidneys that do not adequately function?
2. Explain how the following systems interact with the urinary system.
   - **a** Circulatory
   - **b** Nervous
   - **c** Digestive
   - **d** Respiratory
3. When blood is present in urine, it can be an indication of several disorders, such as high blood pressure. How could high blood pressure be responsible for these results?
4. Explain the interaction that occurs between the circulatory system and kidneys to create urine.

9780170449090

CHAPTER 7 SUMMARY

- Excretion is the removal of the waste products of metabolism.
- The lungs, liver, sweat glands, alimentary canal and kidneys are involved in excretion.
- Deamination is the removal of the amino group from amino acids so that the remaining part can form carbohydrates to be used for energy production. This occurs in the liver.
- The amino group is converted to ammonia and then to urea to be excreted by the kidneys.
- The skin covers the body, helps to regulate the body temperature and is involved in the excretion of salt, lactic acid, urea and some drugs through sweat.
- The kidney, bladder, urethra and ureters make up the urinary system.
- The renal capsule surrounds the outer renal cortex, which lies outside the renal medulla.
- The renal medulla is divided into renal pyramids by the renal columns.
- The renal pelvis is a hollow that leads to the hilum on the concave side of the kidney.
- The nephron is the microscopic unit of the kidney and is made up of a renal corpuscle and a renal tubule.
- Blood is filtered in the renal corpuscle as the blood travels through the glomerulus. The filtrate is collected in the glomerular capsule.
- Selective reabsorption of useful substances including water, glucose, amino acids and some ions occurs in the proximal convoluted tubule, Loop of Henle, distal convoluted tubule and collecting duct.
- Tubular secretion moves substances such as drugs, potassium, creatinine and hydrogen ions from the blood to the urine in the distal convoluted tubule.
- Urine travels from the collecting ducts to the renal pelvis and then the ureters. It then travels to the bladder where it is stored before excretion through the urethra.
- In a healthy person, urine is composed of water, ions, urea, uric acid and creatinine.
- Kidney stones are solid crystals formed when the urine is too concentrated. These may block the ureter, bladder or urethra, causing pain.
- Kidney failure means that the blood is not able to be filtered correctly. Lifestyle choices such as diet, smoking, alcohol consumption and drugs can affect the health of the kidneys.
- Dialysis, either peritoneal or haemodialysis, is used to remove wastes from the blood of people with kidney failure.
- Liver disease, caused by infection, autoimmune disease, genetic disorders, cancer and lifestyle choices, means that toxins are unable to be processed for elimination.

9780170449090

CHAPTER 7 GLOSSARY

Afferent arteriole The blood vessel that enters an organ

Collecting duct The tube in the kidney that collects filtrate from several nephrons

Creatinine A waste product from the breakdown of muscles

Deamination The removal of the amino group from an amino acid molecule

Dialysis A method of removing waste from the blood when kidney failure occurs

Distal convoluted tubule The second set of convolutions of the kidney tubule; it receives the forming urine after it has passed through the loop of Henle

Efferent arteriole The blood vessel that leaves a glomerulus in the kidney

Excretion Removal of the wastes of metabolism from the body

Facultative reabsorption The process whereby carrier proteins assist the movement of substances through the cell membrane

Filtrate The fluid remaining after filtration has taken place

Glomerular capsule The double-walled cup-like structure at the end of each kidney tubule; it collects filtered water and other substances from the blood; also called Bowman's capsule

Glomerular filtration The filtration of blood in the kidney

Glomerulus The tightly coiled mass of capillaries that is surrounded by the expanded part of each kidney tubule

Haemodialysis Process in which blood is passed through an artificial kidney or dialysis machine

Kidney failure Occurs when the kidneys lose their ability to excrete wastes and control the level of fluid in the body

Kidney stone Solid masses of crystals that develop within the kidneys

Kidneys The principal excretory organs of the human body; they filter waste from the blood and regulate the balance of water and salts in the blood plasma

Liver Organ that is part of the excretory system

Loop of Henle The U-shaped section of the kidney tubule; it plays a major role in the reabsorption of water and salts from the filtrate

Lung One of a pair of organs for gas exchange, occupying the chest cavity

Nephron The functional unit of the kidney

Peritoneal dialysis Dialysis occurring inside the body, using the peritoneum as the membrane across which wastes can be removed

Peritoneum The membrane that lines the inside of the abdominal cavity

Peritubular capillaries The capillaries in the kidney that surround the convoluted tubules of the nephron, the loop of Henle and the collecting duct

Proximal convoluted tubule The first set of convolutions of the kidney tubule, located between the glomerular capsule and the loop of Henle

Renal artery The blood vessel that transports blood into the kidney

Renal capsule A tough fibrous layer surrounding the kidney

Renal column An extension of the renal cortex that divides the renal medulla into renal pyramids

Renal corpuscle The filtration structure of the nephron composed of the glomerulus and glomerular capsule

Renal cortex The outer part of the kidney

Renal hilum A depression in the kidney where the blood vessels and the ureter enter

Renal medulla The inner part of the kidney

Renal pelvis The cavity of the kidney that collects urine before it passes to the ureter

Renal pyramid A section of the renal medulla

Renal tubule The kidney tubule; it leads away from the glomerular capsule and empties into a collecting duct

9780170449090

Renal vein The blood vessel transporting blood away from the kidney

Secretion by the tubules *see* tubular secretion

Selective reabsorption The reabsorption of some substances and not others in the renal tubules

Sweat glands Tubular structures in the skin that produce sweat

Tubular secretion The process whereby ions and drugs are secreted from the blood into the kidney tubule

Ureter The tube that leaves each kidney and drains into the urinary bladder

Uric acid A breakdown product of the metabolism of purines such as the bases adenine and guanine found in DNA and RNA

Urinary system The system composed of the kidneys, ureters, bladder and urethra that is responsible for the excretion of wastes, the regulation of blood volume, pressure and pH, as well as the concentration of chemicals

Urine The fluid produced by the kidneys that contains wastes and excess materials

9780170449090

CHAPTER 7 REVIEW QUESTIONS

Recall

1 a What is meant by the term 'excretion'?
  b Which organs of the body are involved in excretion?
2 a Draw a nephron and its associated blood vessels. Label the afferent and efferent arterioles, glomerulus, glomerular capsule, distal and proximal convoluted tubules, loop of Henle, collecting duct and peritubular capillaries.
  b Use arrows on your diagram to indicate the direction of blood flow and the direction in which the filtrate flows.
3 a Define 'deamination'.
  b Describe where deamination occurs.
  c Describe what happens to the ammonia produced in deamination.
4 Describe the role of the skin in excretion.
5 Describe what happens in the nephron during:
  a filtration
  b reabsorption
  c secretion.
6 Describe kidney failure.

Explain

7 Explain why ammonia must not accumulate in the tissues.
8 a Explain the process of dialysis.
  b Explain the difference between peritoneal dialysis and haemodialysis.
9 Make a list of ways in which the structure of the kidney is suited to the functions that it performs. For each structural feature on your list, explain how that feature is related to the working of the kidney.

Apply

10 Compare and contrast the filtrate and the blood entering the glomerulus.
11 What lifestyle measures should you adopt to make sure that your kidneys remain healthy?
12 To be effective, an organ where materials are taken into the body, or passed out of the body, must have a very large surface area. How is a large surface area achieved in the kidney?
13 In this chapter, you were told the approximate length of a renal tubule and also the approximate number of tubules in a kidney. Using these figures, calculate the total tubule length for an average person, remembering that most people have two kidneys. Express your answer in appropriate units.
14 The kidneys have a very important function in maintaining the composition of body fluids at a constant level. Describe the kidney's role in maintaining a constant internal environment for the cells.
15 What effects would you expect the following to have on urine production?
  a A high-salt diet
  b A low-protein diet
  c A large intake of water
16 a Suggest why we tend to urinate more frequently in cold weather than in hot weather.
  b Suggest why urine is often more darkly coloured in hot weather than in cold weather.

9780170449090

Extend

17 The desert hopping mouse lives on dry seeds and never drinks water. It has extremely long kidney tubules. How is it able to reduce water loss in urine to a minimum?

18 Explain how the circulatory system forms a link between the respiratory, digestive and excretory systems.

19 Why do doctors sometimes order a urine test for a patient? What information about a person's health can be gained from an analysis of their urine?

20 Kidney disease causes protein to be excreted in the urine. The loss of protein from the blood causes fluid to accumulate in the tissues and there may be swelling of the hands, feet and face. Conduct research to find out why loss of protein from the blood causes swelling of the tissues.

21 Countercurrent exists between the two limbs of the loop of Henle and between each nephron tubule and surrounding capillaries. Conduct research to find out:

- **a** what a countercurrent is
- **b** the importance of the countercurrents that exist in the kidney
- **c** how changes in the concentration of the filtrate would affect the countercurrent exchange mechanisms
- **d** other places in the body, apart from the kidney, where countercurrent exchanges operate.

9780170449090

8 THE MUSCULO-SKELETAL SYSTEM ALLOWS MOVEMENT

UNIT 1 CONTENT

SCIENCE INQUIRY SKILLS

» conduct investigations, including monitoring body functions; use microscopy techniques; and perform real or virtual dissection, safely, competently and methodically for the collection of valid and reliable data

SCIENCE AS A HUMAN ENDEAVOUR

» osteoporosis and osteoarthritis are diseases, primarily of ageing, that cause disability. Increased understanding of the causes of these conditions leads to improved practices for management and prevention

SCIENCE UNDERSTANDING

Musculoskeletal system

» the muscular system is organised to maintain posture and produce movement; muscle fibre contraction can be explained using the sliding filament theory

» movement results from the actions of paired muscles, with others acting as stabilisers, to produce the required movement

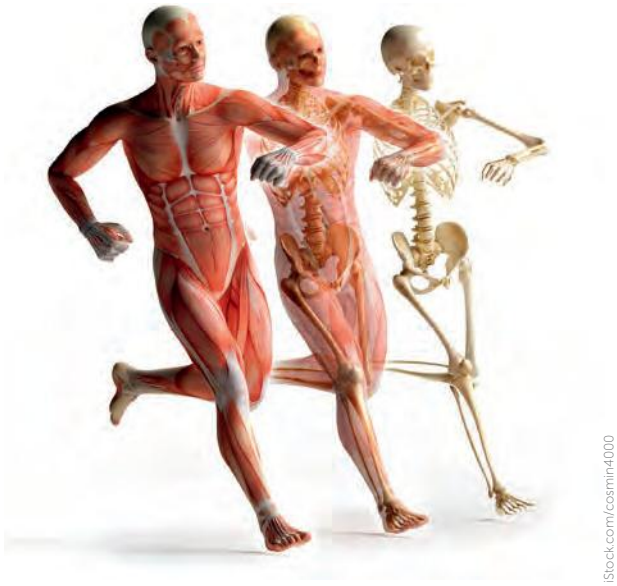
» the skeletal framework of the body consists of bone and cartilage which function to provide body support, protection and movement, and is facilitated by the structure and function at cell and tissue levels

» articulations of joints of the skeleton are classified according to their structure or the range of movements permitted

Source: School Curriculum and Standards Authority, Government of Western Australia

9780170449090

The musculoskeletal system is composed of the skeletal system and the muscular system. These work together to support and move the body.



iStock.com/cosmin4000

FIGURE 8.1 The muscular and skeletal systems work together

Science Photo Library/Gustoimages

FIGURE 8.2 The muscles move the bones, allowing movement

As we saw in Chapter 2, there are four basic types of tissue: epithelial, connective, muscular and nervous tissue. Muscle cells are in the form of long, thin fibres that have the ability to **contract**, or shorten. This characteristic distinguishes muscle from all other body tissues. Muscles are organised in such a way that, when they contract, they reduce the distance between the parts they are connected to, or decrease the space they surround. Bone, cartilage, tendons and ligaments are examples of connective tissue and work with muscles to create movement. These tissues are characterised by the cells being separated by non-cellular matrix.

The skeleton gives shape and form to the body and prevents the soft tissues from collapsing in a heap. Muscles attached to the bones are able to move them or hold them steady, enabling us to stand erect, walk, run, jump and perform complicated movements like the one shown in Figure 8.2.

8.1 TYPES OF MUSCLES

Muscle tissue
This website provides a brief review of the types of muscle.

There are three types of muscle: skeletal, smooth and cardiac muscle. The muscles that move bones and enable us to walk, run and carry out a wide range of voluntary physical activities are the **skeletal muscles**. These muscles are under conscious control and are attached to the bones of the skeleton. Contractions of the skeletal muscles bring about movement at the joints. They also give the body its form and contours, and allow it to maintain posture.

Many of the internal organs, such as the stomach and intestines, have muscles for movement. These are known as **smooth muscles** or, because they are not under conscious control, **involuntary muscles**. When the smooth muscles that wrap around the alimentary canal contract, the diameter of the canal narrows, pushing the contents along.

A third type of muscle is heart muscle. This is known as **cardiac muscle**. When cardiac muscle contracts, it reduces the space in the chambers of the heart and pushes the blood from the heart into the blood vessels.

Apart from the ability to contract, muscles possess the properties of extensibility and elasticity. **Extensibility** is the ability to be stretched, while **elasticity** is the ability to return to the original length after being stretched. These three properties – contractibility, extensibility and elasticity – allow muscles to work together to create movement. It is important to note that, while muscles are able to shorten their length and can be stretched, they are unable to increase their own length. The remainder of this chapter concentrates on the muscles associated with the skeletal system and how they work together to bring about movement at a joint.

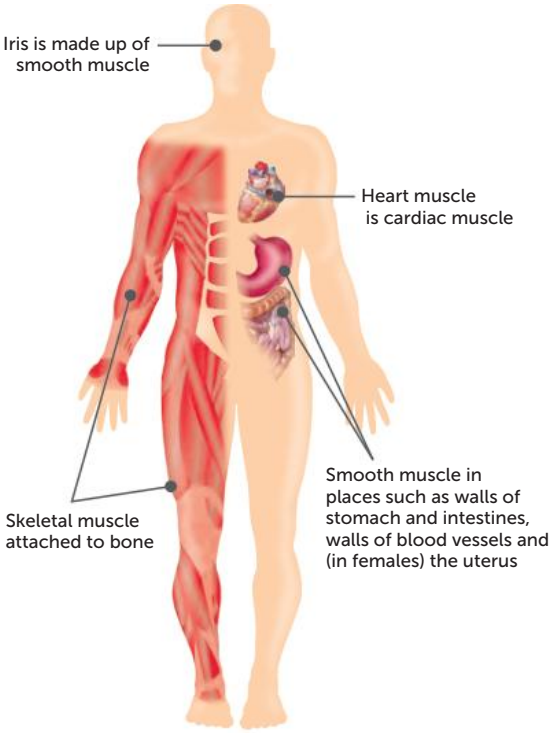


FIGURE 8.3
Locations of the three types of muscle tissue

Questions 8.1

RECALL KNOWLEDGE

1 List the types of muscle cells.
2 Describe the properties of muscles.
3 Which types of muscles are under involuntary control?

APPLY KNOWLEDGE

4 List the type of muscle that is associated with each of the following:
   a the heart
   b the oesophagus
   c the hand
   d the left ventricle
   e the lower leg
   f the small intestine.
5 Predict the consequence of a muscle losing its elasticity.

Muscle names
This website provides the location and names of individual muscles.

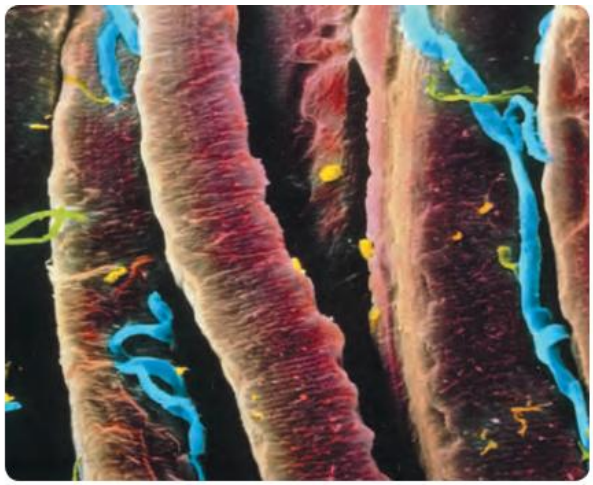
8.2 STRUCTURE OF SKELETAL MUSCLE

Muscle cells are held together in bundles. A sheath of connective tissue called the perimysium surrounds each bundle so that it can function as an individual unit. The connective tissue allows adjacent bundles to slide easily over one another as they contract. Sheaths of connective tissue called epimysium also hold the bundles together, and towards the end of the muscle they taper and blend to form the tendon.

The amount of connective tissue increases with advancing age. Increased amounts of connective tissue are also thought to contribute to the decrease in muscular strength that gradually occurs as a person gets older.

Red meat is muscle. It is the bundles of muscle cells that give meat its 'stringy' appearance when cut lengthwise, while the connective tissue gives the meat a certain toughness. Meat from an older animal is less tender due to the increase in connective tissue.

9780170449090



Science Photo Library/Prof. P Motta/Dept. of Anatomy/University La Spienz, Rome

FIGURE 8.4 Coloured electron micrograph of the skeletal muscle. The muscle bundles are shown in brown and the capillaries in blue

Structure of skeletal muscle fibres

If one of the muscle bundles is examined under a microscope, it is possible to see that it is composed of muscle cells that lie parallel to each other. Each muscle cell, called a **muscle fibre**, is an elongated cylinder with many nuclei. Around the cell is a thin, transparent plasma membrane, the **sarcolemma**, containing cytoplasm, called the **sarcoplasm**. Cells are between 10 and 100 micrometres in diameter and vary in length from a few millimetres to several centimetres.

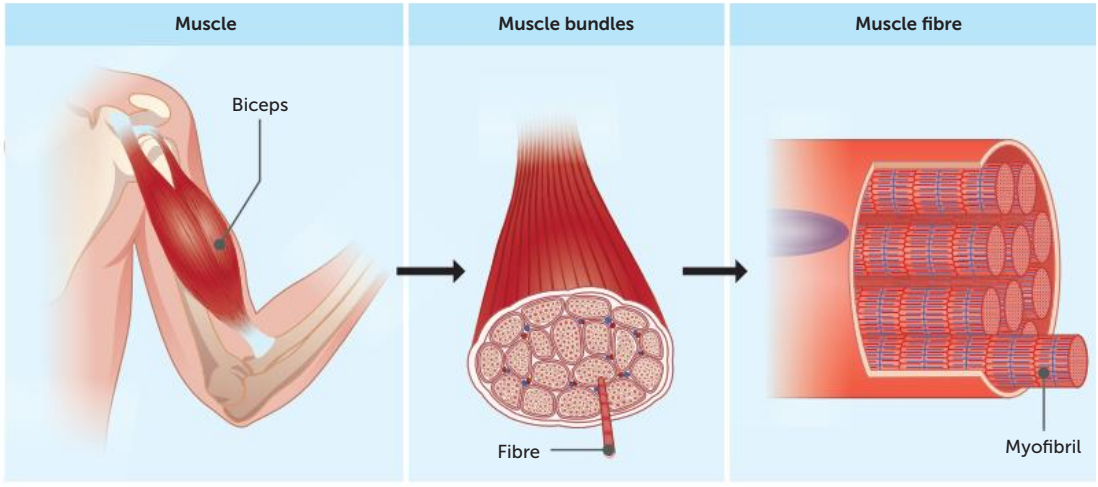


FIGURE 8.5 The relationship between muscles, muscle bundles and muscle fibres

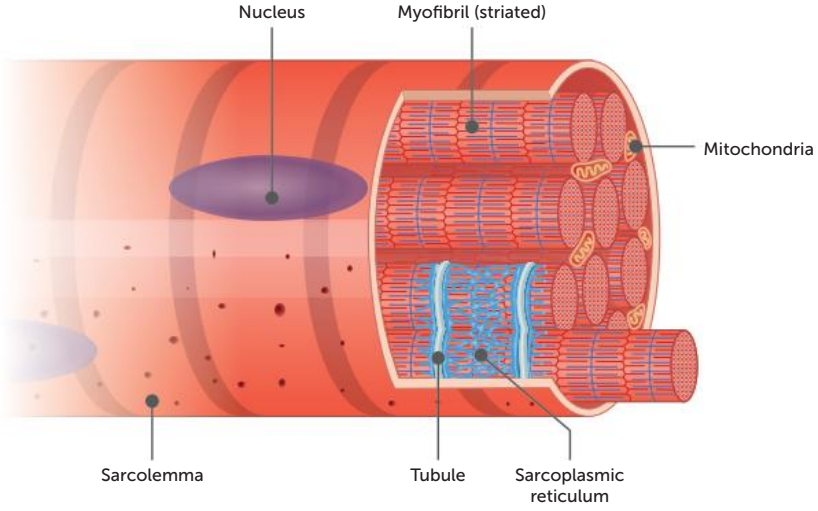


FIGURE 8.6 Structure of a muscle fibre

Structure of myofibrils

Within the sarcoplasm of each fibre are thread-like **myofibrils**. These lie parallel to each other and run the length of the fibre. There may be anywhere from hundreds to several thousands of these myofibrils in each fibre. A tubular network called the sarcoplasmic reticulum surrounds the myofibrils. This is a storage site for calcium ions, which are released during muscle contractions.

Each myofibril is composed of many smaller **myofilaments**, made of protein, which are the actual units involved in contraction of the muscle. There are two types of myofilaments:

- thick myofilaments, composed mainly of the protein **myosin**
- thin myofilaments, composed mainly of the protein **actin**.

When a muscle fibre is supplied with sufficient energy, in the form of adenosine triphosphate (ATP), and is activated by a nerve impulse, these thick and thin protein filaments slide past each other in a manner that shortens the myofibril.

The arrangement of thick and thin filaments within a myofibril gives a banded effect to the muscle. It is these bands that give skeletal and cardiac muscle fibres their striated appearance when viewed under a microscope (see Figure 8.8). These striations allow myofibrils to be divided into units called **sarcomeres**.

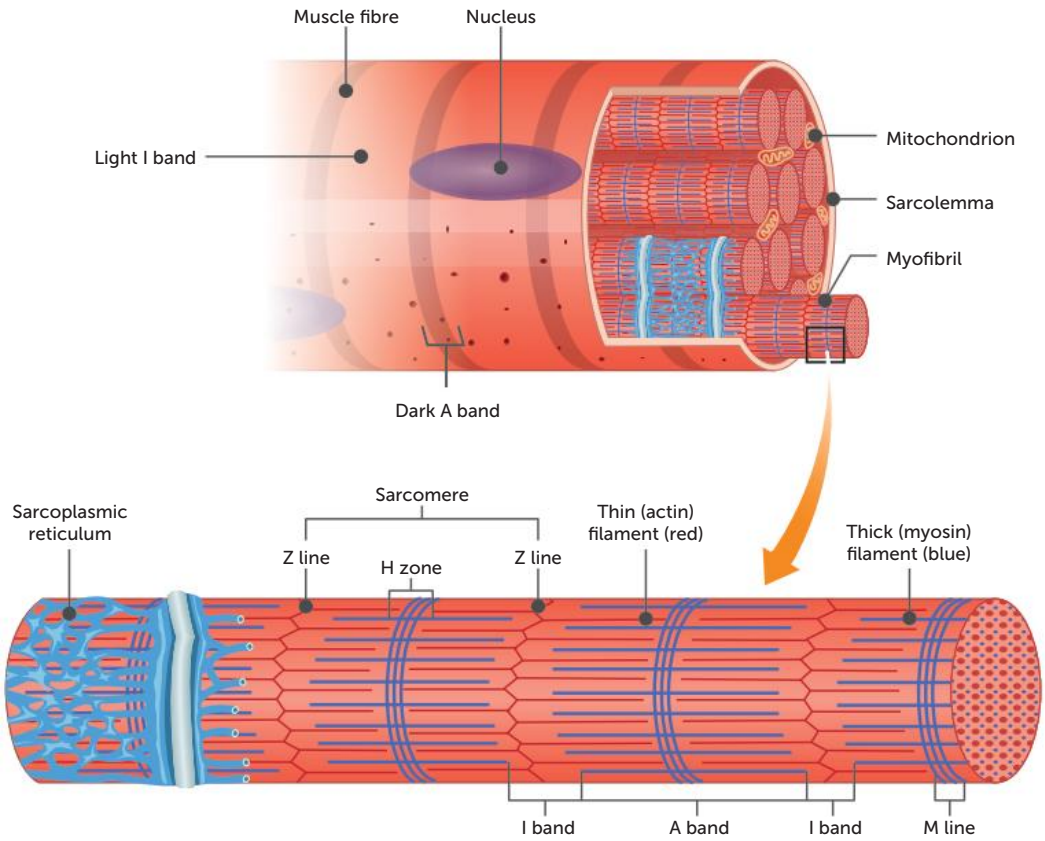


FIGURE 8.7 The relationship between a muscle fibre, myofibril and myofilament

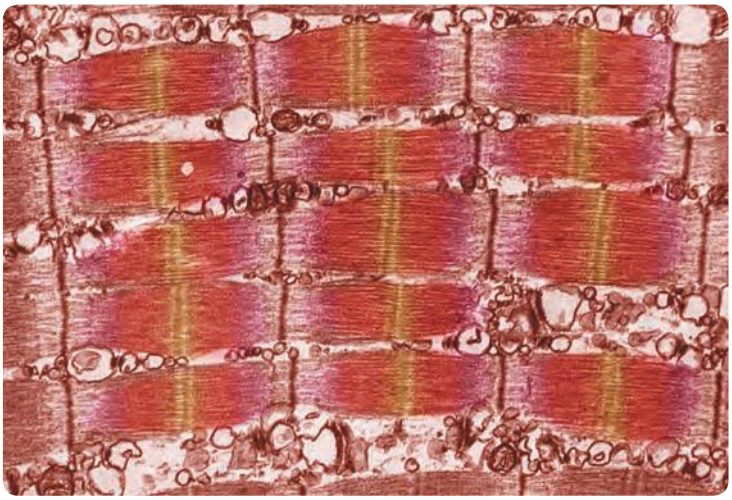


FIGURE 8.8 Electron micrograph of skeletal muscle showing sarcomeres

Key concept

Skeletal muscle is made of bundles of muscle fibres that contain myofibrils, which contain myofilaments made of the proteins actin and myosin.

Questions 8.2

RECALL KNOWLEDGE

1. What holds bundles of muscle cells together?
2. Place the structures in order from the smallest to the largest: actin and myosin, muscle, myofilament, muscle bundle, myofibril, muscle fibres.
3. Define 'sarcolemma'.

APPLY KNOWLEDGE

4. Explain why meat from younger animals is of a higher quality than meat from older animals.
5. Explain why skeletal muscle has a striated appearance.

8.3 HOW MUSCLES WORK

Muscles have the properties of excitability, contractibility, extensibility and elasticity. This means that they are able to:

- be stimulated by a nerve impulse
- shorten in length
- be stretched
- return to their original length.

Sliding filament theory

Muscles are the only tissues that have the ability to contract. To explain how muscle contraction occurs, a **sliding filament theory** has been proposed. When muscles contract, the sarcomeres shorten. The theory suggests that this occurs because the actin and myosin filaments slide over one another.

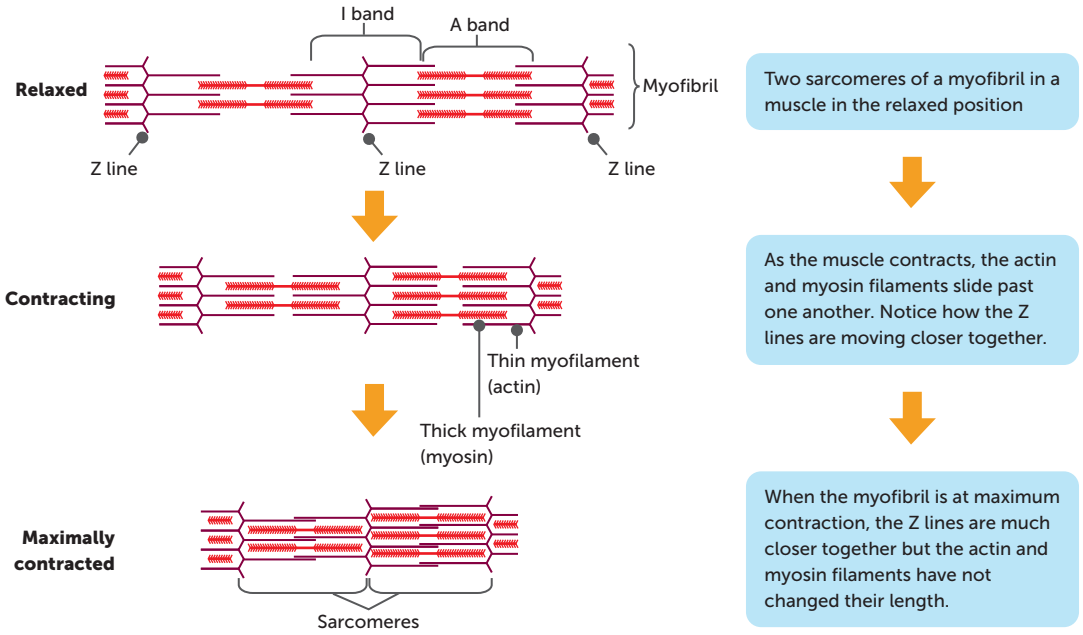


FIGURE 8.9 Sliding filament theory of muscle contraction: the various parts of two sarcomeres are shown in relaxed, contracting and maximally contracted positions

In the sliding filament theory, the middle of the thin filaments contains protein discs called Z lines. The distance between successive Z lines is a sarcomere. The length of the thick filament (myosin) is called the A band. At the ends of the A band, the thin and thick filaments overlap. The middle of the A band is lighter, as it contains the thick filaments only. This region is called the H zone. The distance between successive thick filaments is called the I band, which contains only thin filaments. As the thin actin filaments slide over the thick myosin filaments, the Z lines are drawn closer together and the sarcomere is shortened. This results in a shortening of the muscle fibres and, hence, a shortening of the whole muscle.

As you can see in Figure 8.9, the myofilaments are the same length in the contracted position as they were before contraction. The fibril has shortened because the myofilaments overlap more. When the muscle relaxes, the actin and myosin filaments can be pulled past one another in the opposite direction and the muscle fibre returns to its original, uncontracted state. At any given time, some muscle fibres can be contracted, while others are relaxed.

Energy is required for shortening of the muscle fibres. The energy comes from the breakdown of ATP in the muscle cells (see Chapter 3). Energy is released when ATP breaks down to adenosine diphosphate (ADP) and a phosphate group. When energy is again available from cellular respiration, ATP is re-formed. ATP is the molecule that transfers energy from cellular respiration to processes such as muscle contraction.

Activity 8.1
Investigating fast- and slow-twitch fibres

Video of sliding filaments
This website has a short video showing an animation of the sliding filament model.

Sliding filament theory of muscle contraction
This website includes an animation of the sliding filament theory.

Key concept

The sliding filament theory is used to explain what happens during muscle contraction, with actin and myosin filaments moving over one another and shortening the length of the muscle.

Skeletal muscles working together

Muscles are attached to the bones of the skeleton by fibrous, inelastic connective tissue called **tendons**. They are attached to the bones in such a way that they bridge the joints (Figure 8.10), so when muscle contraction occurs the bones move.

Muscles can only contract. This means that they can pull bones together, but they cannot push them apart. If muscles contract, pulling a bone in one direction, another set of muscles must contract to pull the bone in the opposite direction. Thus, the muscles that move the parts of the skeleton are always grouped in pairs.

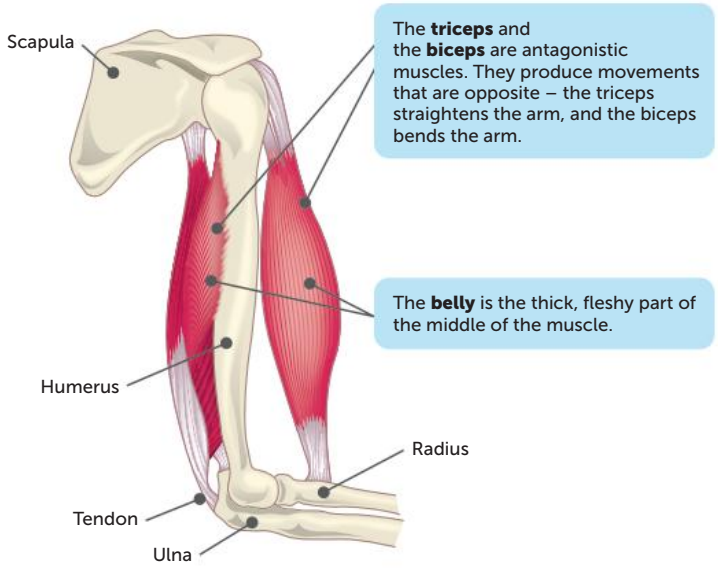


FIGURE 8.10 Muscles work in pairs

Coordination of the paired muscles provides body movement, with one of the pair producing movement of bones in one direction and the other producing movement in the opposite direction. Such pairs of muscles are referred to as **antagonists**, as they have opposite actions. The muscles of the upper arm are good examples of antagonists. As Figure 8.10 shows, one end of the muscle on the front part of the upper arm, the **biceps**, is fixed to the scapula (shoulder blade). The other end of the biceps muscle is attached to the radius. The muscle at the back of the upper arm, the **triceps**, is fixed to the scapula and to the humerus at one end, and to the ulna at the other. To move the forearm about the elbow joint, these two muscles must cooperate. When the biceps muscle contracts to bend the arm, the triceps must relax; the opposite occurs when the arm is straightened.

The hamstring and quadriceps muscles in the legs are also an antagonistic pair.

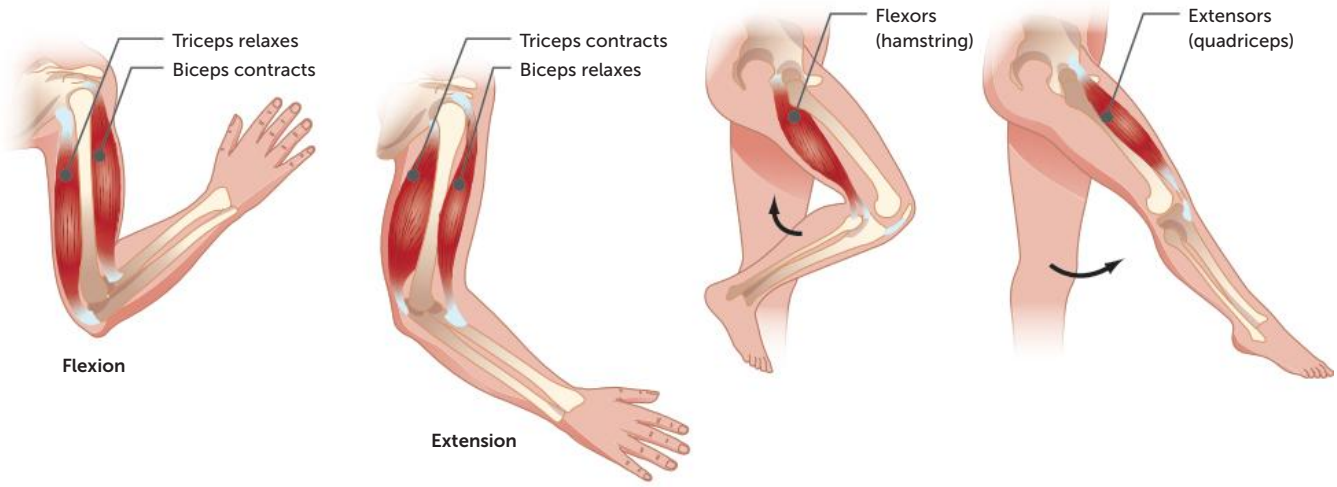


FIGURE 8.11 Antagonistic muscles work in opposite ways

When the biceps muscle contracts, the shoulder end remains stationary, but the other end moves and bends the forearm. The end of the muscle fixed to the stationary bone is called the **origin**. The attachment to the movable bone of the other end of the muscle is the **insertion**. The fleshy portion of the muscle between the tendons of the origin and the insertion is called the **belly**. A muscle that causes a desired action is called the **agonist**, or **prime mover**. As mentioned, while the biceps is contracting, the triceps is relaxing. In this situation the biceps is the agonist, because it moves the forearm; the triceps is the antagonist, because it has an effect that is opposite to that of the agonist – that is, it yields to the movement of the agonist. However, when the triceps is contracting to straighten the forearm, the biceps is relaxing and their roles are reversed. Now the triceps is the agonist and the biceps the antagonist.

Muscles working as pairs
This BBC website provides a view of the biceps and triceps working together.

9780170449090

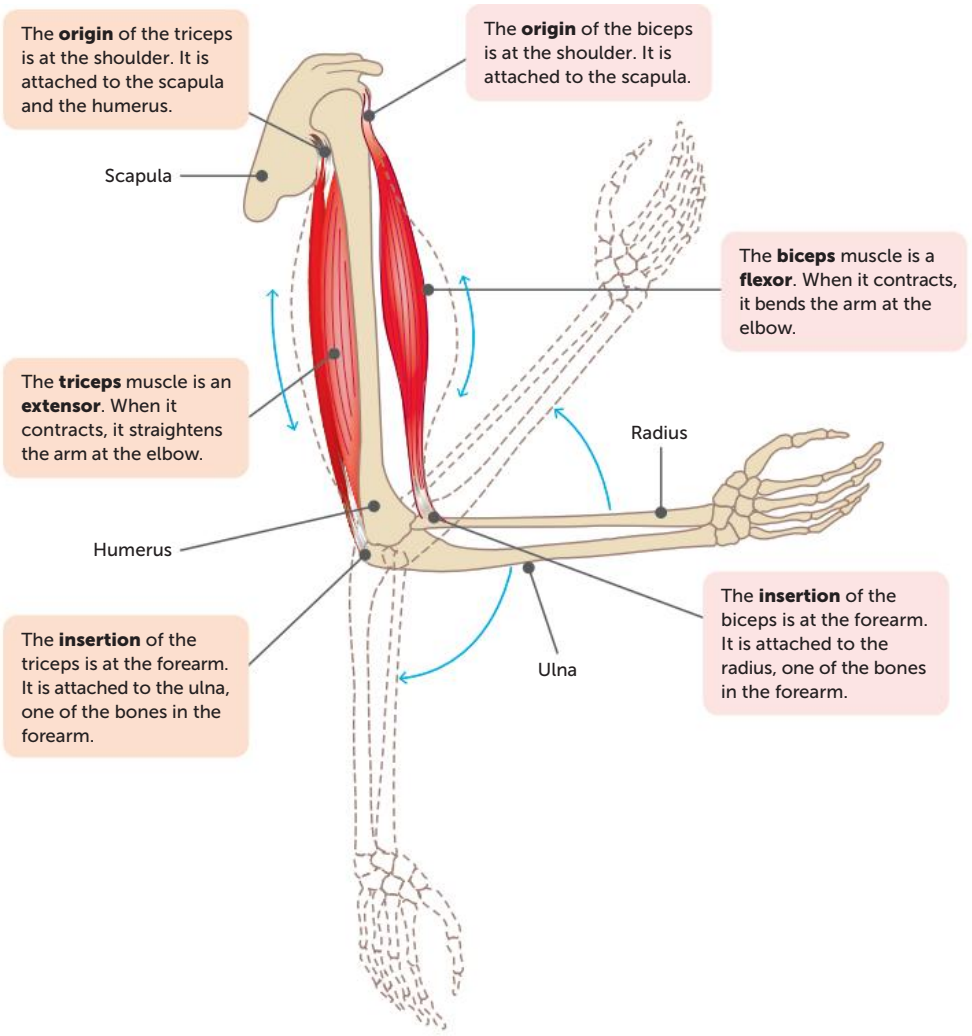


FIGURE 8.12 The origin and insertion of the triceps and biceps – an antagonistic pair

Key concept

Muscles work in pairs, with the muscles fulfilling opposite roles.

In addition to agonists and antagonists, most joint movements involve muscles that act as synergists. **Synergists** are muscles that help the prime mover. They may produce the same movement as the prime mover, or they may steady a joint during a particular movement so that unwanted movement is prevented and the prime mover (agonist) can function more efficiently. For example, the wrist would flex every time the fist was clenched if it were not for synergistic muscles, because the muscles that curl the fingers also pass across the wrist. Synergistic muscles immobilise the wrist, stopping it from flexing.

When a synergist immobilises a joint in this way, it is called a **fixator**. A fixator muscle acts as a stabiliser of one part of the body during movement of another part. Other examples of fixators are the many muscles that attach the scapula to the axial skeleton. These are important as fixators because the scapula is only attached to the axial skeleton at the ribs. To act as a firm origin for the muscles that move the arm, it must be held steady when those muscles contract. The fixators hold the scapula firmly against the chest (or thorax), so when the arm muscles contract, only their insertions are moved. The interaction of agonists, antagonists and synergists makes very fine and precise movements possible.

A muscle may assume different roles depending on the body movement that is occurring. The same muscle may at different times be an agonist, an antagonist or a synergist.

WS

8.1 Working muscles

9780170449090

Muscle tone

Muscle tone is maintaining partial contraction of skeletal muscles. At any one time, some muscle fibres are contracted while others are relaxed. Such partial contraction tightens a muscle, but not enough fibres are contracting at the one time to produce movement. Muscle tone is not caused by the constant contraction of the same fibres, but by many different fibres taking turns to contract. The fibres relieve one another so smoothly that the contraction can be kept up for long periods of time.

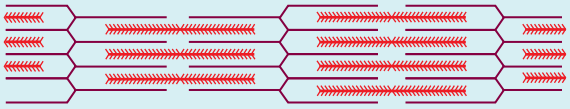
Muscle tone holds many of our body parts in position. For example, the head is held up by the partial contraction of the neck muscles. When a person falls asleep in a chair, their head droops. This is due to loss of tone in the neck muscles.

A person's **posture** is the characteristic way they hold their body when standing or sitting. It is the way a person positions their body or arranges their limbs. A ballerina, for example, may have an elegant, graceful posture, while a soldier may appear more rigid. Posture depends on muscle tone – that is, the partial contraction of those muscles that hold the body in position.

Questions 8.3

RECALL KNOWLEDGE

1. List the properties of muscles.
2. Define 'sarcomere'.
3. Classify actin and myosin as muscle fibres, myofibrils or myofilaments.
4. Clearly identify each of the following on the diagram below.
   Z line, A band, I band, actin, myosin, H zone, sarcomere



5. Use the hamstring and quadriceps as an example to explain the difference between an agonist and antagonist when the leg is flexed.
6. Describe the importance of synergists.

APPLY KNOWLEDGE

7. Explain what happens to each of the following when a muscle contracts.
   a The A band
   b The I band
   c The sarcomere
8. Predict what would happen if the tendon attaching the triceps muscle to the bone were severed.

8.4 OVERVIEW OF THE SKELETAL SYSTEM

The skeletal system is made up of the bones and their associated structures: tendons, ligaments and joints.

Functions of the skeleton

The skeleton is much more than simply a rigid framework that gives shape to the body.

- It provides a scaffold to support the weight of the rest of the body.
- It facilitates movement by being points of attachment for muscles. When muscles contract, they allow movement to take place. Where bones meet so that they are able to move relative to one another, they are said to articulate. **Articulation** allows varying degrees of movement, depending on the bones involved, so the skeleton determines the extent of movement of body parts.

9780170449090

- It protects vital internal organs. For example, the brain is encased within the cranium, the spinal cord is contained within the spinal canal formed by the vertebrae, the heart and lungs are protected by the rib cage, and the pelvis protects the internal reproductive organs and bladder.
- It produces red blood cells. The red marrow contained within certain bones contains stem cells that can differentiate into blood cells. Factors in the cells' environment determine whether they become red blood cells, white blood cells or platelets.
- It stores and releases minerals and fat. The bones of the skeleton act as storage organs for mineral salts and fat. Calcium, phosphorus, sodium and potassium are the main minerals stored within bone. They can be distributed to other regions of the body by the circulatory system when required. For example, if a pregnant woman's diet is lacking in calcium, it can be removed from her skeleton and used for the growth of bones in the developing foetus.

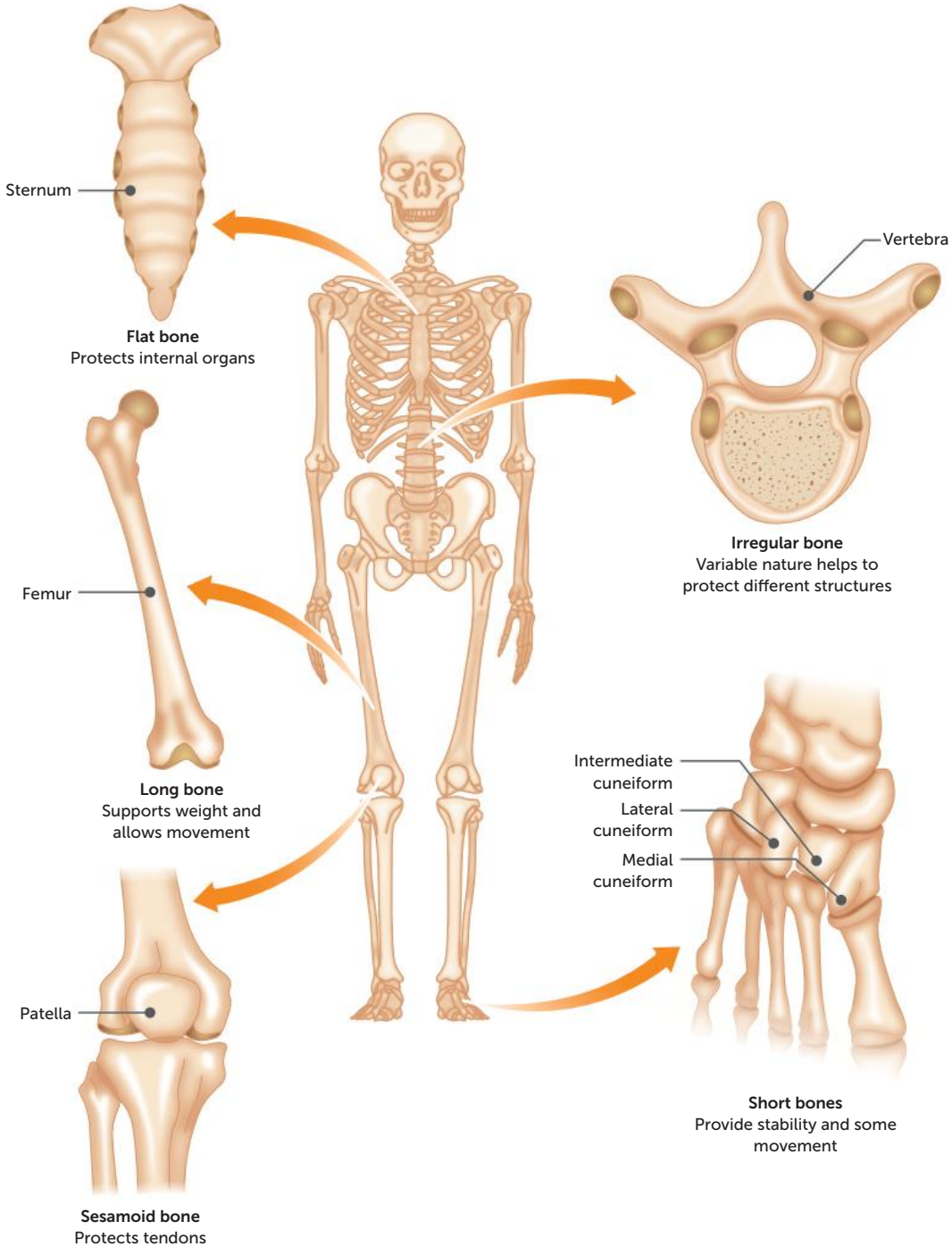


FIGURE 8.13 Types of bone and their functions

9780170449090

Types of bones

Types of bone

There are five different types of bone, based on their shape and structure. The different structures allow each type to fulfil a different function in the body (see Figure 8.13).

Bones of the skeleton

There are normally 206 individual bones making up the adult human skeleton. Some of these bones may be fused to one another to form structures like the skull, but most have joints between them that allow movement.

The bones of the skeleton are divided into two sections.

- The **axial skeleton** consists of the bones that lie around the central axis of the body. It provides the main support for erect posture and protection of the central nervous system and the organs contained within the thorax. The bones that form the skull, vertebral column, ribs and sternum (breastbone) make up the axial skeleton.
- The **appendicular skeleton** consists of the bones of the upper and lower limbs, the pectoral girdle (shoulder) and pelvic (hip) girdle. These two girdles allow for the articulation of the limbs with the axial skeleton.

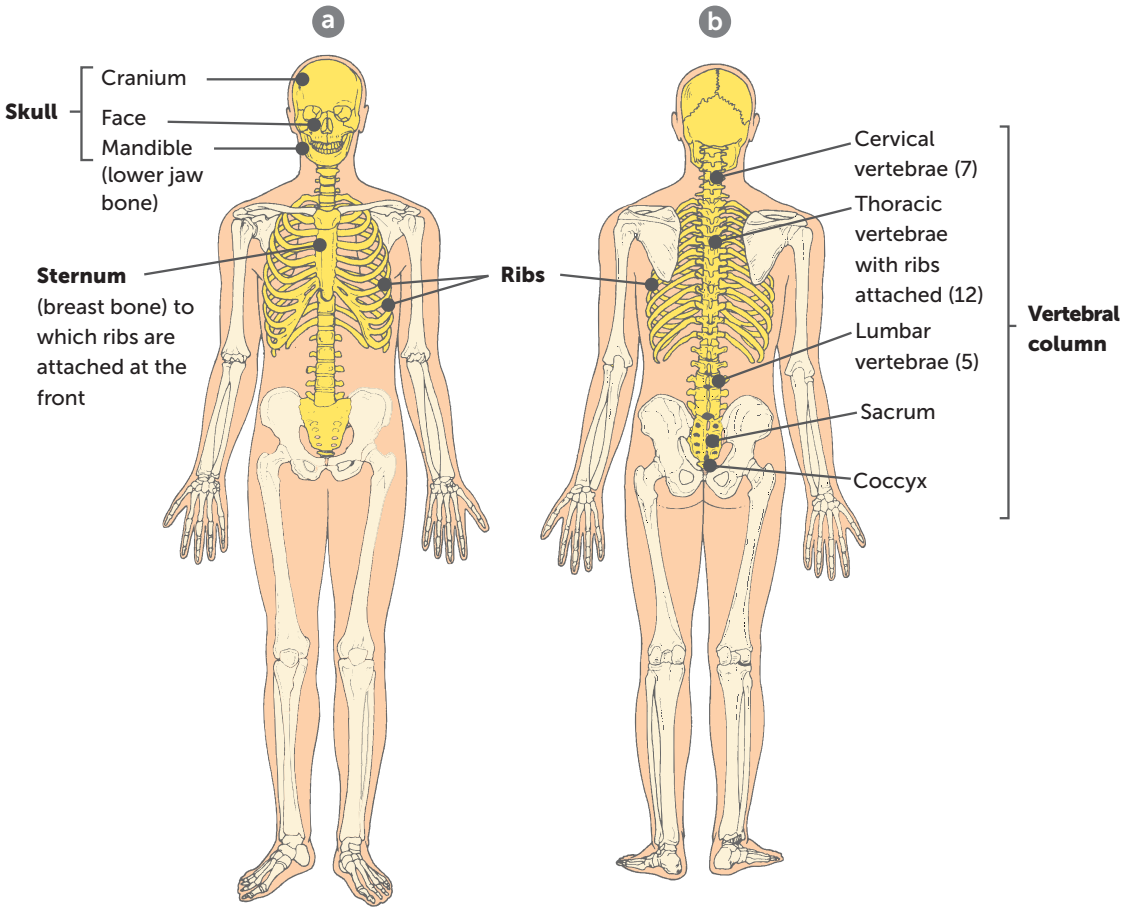


FIGURE 8.14 Axial skeleton: **a** anterior view; **b** posterior view

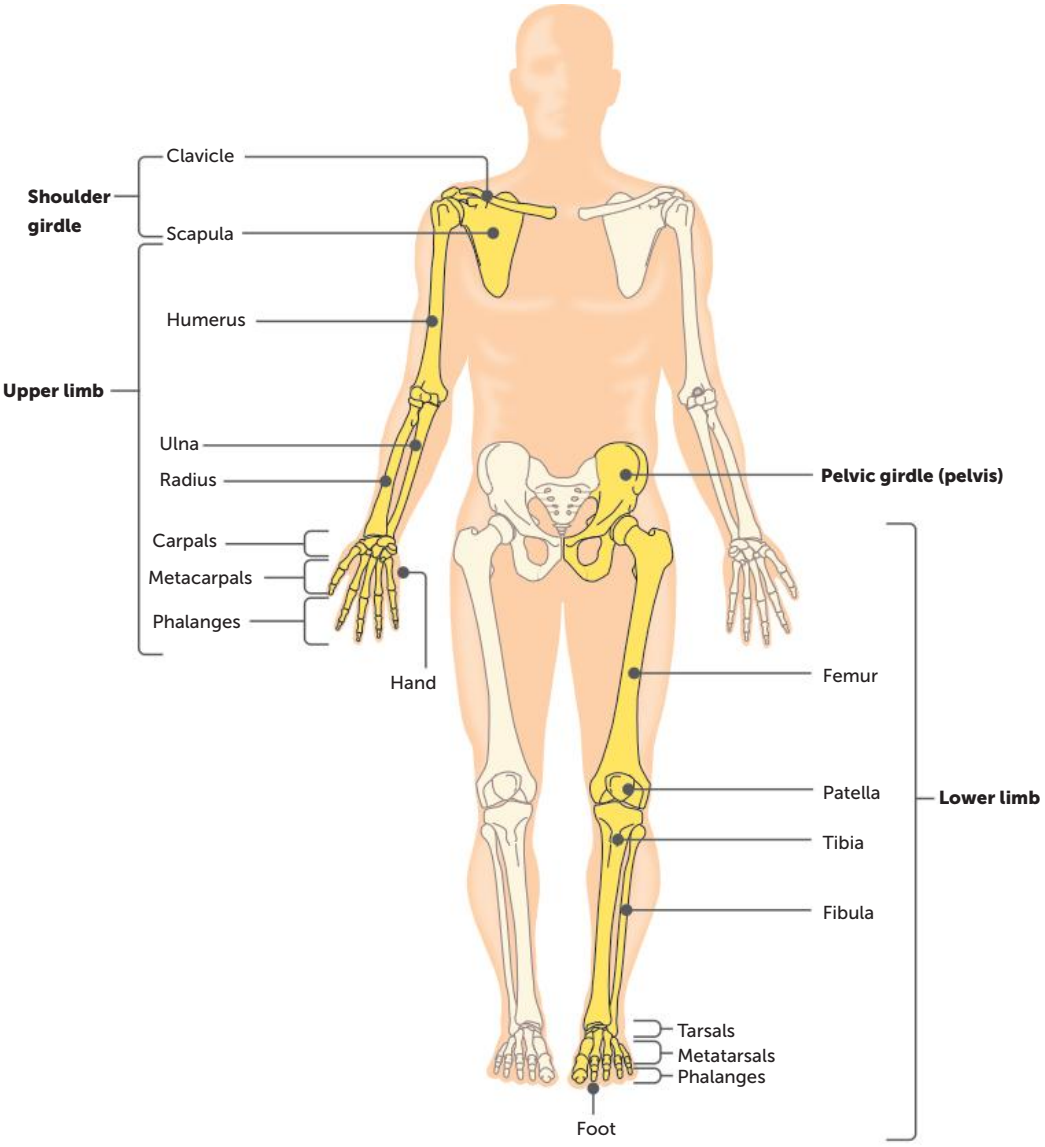
TABLE 8.1 Bones of the skeleton

DIVISION OF THE SKELETON	USUAL NUMBER OF BONES		
Axial skeleton			**80**
1 Skull		29	
• Cranium	8		
• Face	14		
• Hyoid	1		
• Ossicles (ear bones) – malleus, incuse and stapes	6		
2 Vertebral column		26	
• Cervical vertebrae	7		
• Thoracic vertebrae	12		
• Lumbar vertebrae	5		
• Sacrum	1		
• Coccyx	1		
3 Thorax		25	
• Sternum	1		
• Ribs	24		
Appendicular skeleton			**126**
4 Shoulder girdle		4	
• Clavicle	2		
• Scapula	2		
5 Upper limb		60	
• Humerus	2		
• Ulna	2		
• Radius	2		
• Carpals	16		
• Metacarpals	10		
• Phalanges	28		
6 Pelvic girdle		2	
• Coxal bones	2		
7 Lower limb		60	
• Femur	2		
• Fibula	2		
• Tibia	2		
• Patella	2		
• Tarsals	14		
• Metatarsals	10		
• Phalanges	28		
Total			**206**

Bone names
This website provides more information about bones and their scientific names.

9780170449090

FIGURE 8.15
Appendicular skeleton



WS
8.2 The skeleton

The bone game

Key concept

The bones of the axial and appendicular skeletons are classified based on their structure.

Questions 8.4

RECALL KNOWLEDGE

1. List the functions of the skeletal system.
2. State the type of bone for each example.
   a. Cranium
   b. Humerus
   c. Patella
   d. Carpal bones
   e. Pelvis
3. Describe the axial and appendicular skeletons.

APPLY KNOWLEDGE

4. Predict what would happen if the skull consisted of cartilage instead of bone.
5. Explain why it is important for pregnant women to include sufficient calcium in their diet.
6. Suggest why the wrist is composed of short bones.

9780170449090

8.5 STRUCTURE OF BONE AND CARTILAGE

Many people encounter bones only when they see the remains of a dead animal. Therefore, it is no wonder that they might think of bones as a dry, hard, white material. However, bones are actually living organs containing living cells and are capable of growth and repair.

Macroscopic structure of long bones

A long bone consists of the following:

- **Diaphysis** – the shaft making up the main portion of the bone. When the bone is cut lengthwise, the diaphysis is seen to be a hollow cylinder of **compact bone** surrounding a medullary cavity. This cavity is used as a fat storage site and is often called the **yellow bone marrow** cavity.
- **Epiphyses** (singular: **epiphysis**) – the enlarged ends of the bone, covered with a thin layer of cartilage (articular cartilage). The epiphyses have compact bone on the outside, but their central regions contain **spongy** or **cancellous bone**. Cancellous bone is more porous than compact bone, and contains many large spaces filled with marrow. In certain bones, this may be **red bone marrow**, where blood cell production takes place.
- **Periosteum** – the dense, white, fibrous outer covering of the bone. There is no periosteum at the joints, where the bone is covered with an articular cartilage.

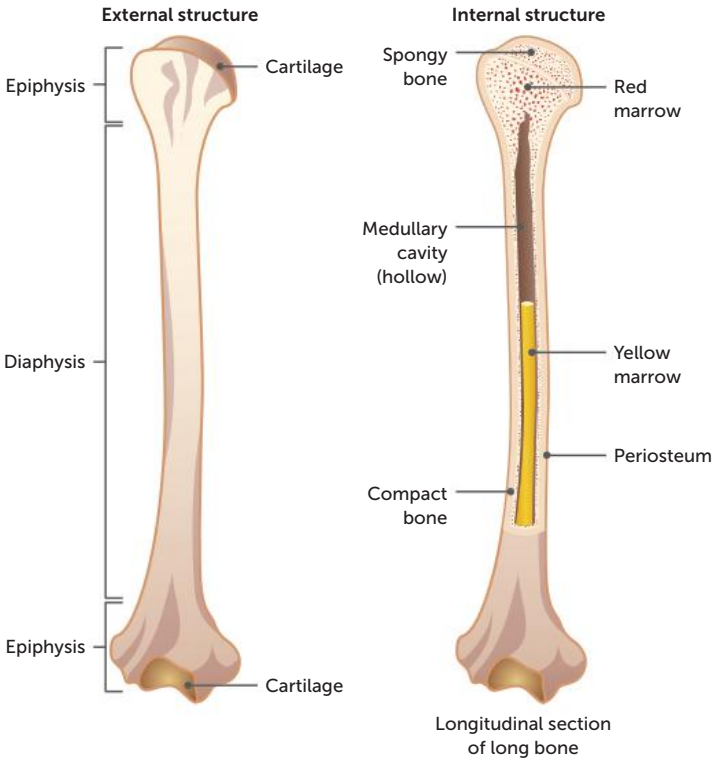


FIGURE 8.16 Macroscopic structure of long bones

Activity 8.2
Studying a long bone

Features of long bones
This website features a graphic that enables you to identify, and gain more information about, the structures in a long bone.

Microscopic structure of bone

In Chapter 2 you learnt that bone is classified as a connective tissue. **Connective tissues** consist of cells separated from each other by large amounts of non-cellular material called **matrix**. In bone, inorganic salts of calcium and phosphate are deposited in the matrix. These increase its rigidity and strength, and make it the hardest of the connective tissues.

When thin slices of bone are examined under a microscope, it can be seen to have a very complex structure.

Activity 8.3
Investigating the composition of bone

9780170449090

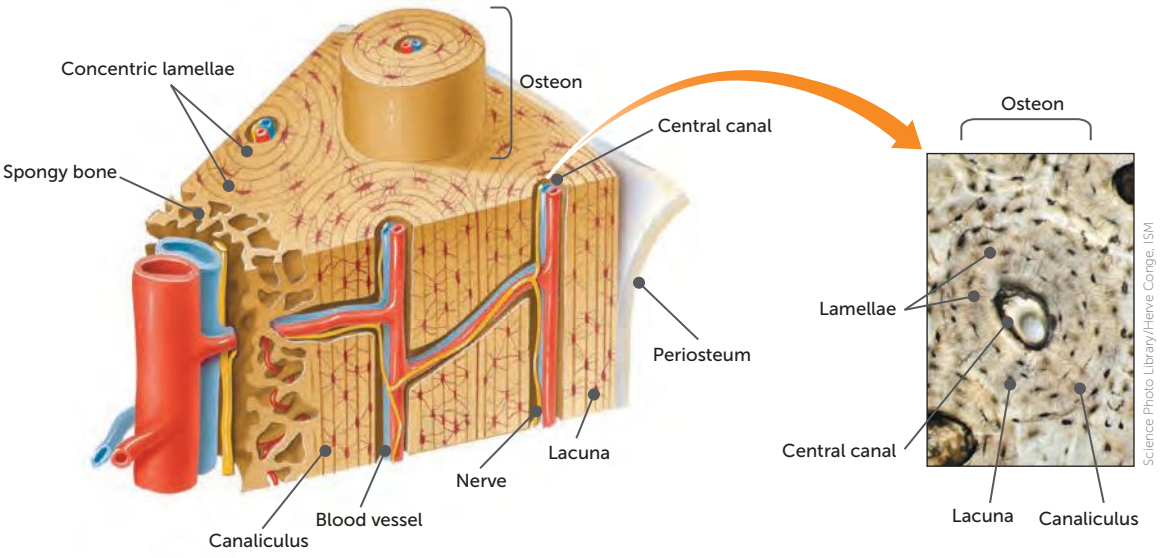
Osteons
This website provides more information on osteons.

Structure of compact bone

Compact bone consists of many similar units called **osteons** or **Haversian systems** (Figure 8.17) that run parallel to the long axis of the bone. This gives the bone its maximum strength. Each osteon has:

- a **central canal** (or **Haversian canal**) at its centre
- concentric layers of bony matrix called **lamellae** surrounding the central canal
- **lacunae**, which are small spaces in the matrix between the lamellae
- a bone cell, or **osteocyte**, occupying each lacuna
- tiny canals, known as **canaliculi**, running between the lacunae
- projections from the bone cells entering the canaliculi and making contact with adjacent bone cells, allowing materials to be passed from cell to cell
- at least one blood capillary, and possibly nerves and lymph capillaries, in the central canal of each osteon.

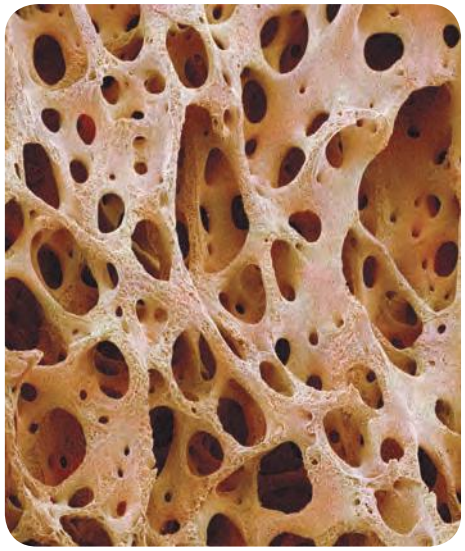
FIGURE 8.17
Microscopic structure of compact bone



Structure of spongy bone

Spongy bone, also called cancellous bone, is not organised into osteons with concentric layers of lamellae. Instead, it consists of an irregular arrangement of thin, bony plates called **trabeculae**. The bone cells occupy spaces in the trabeculae, and nerves and blood vessels pass through irregular spaces in the matrix.

FIGURE 8.18
Scanning electron micrograph of spongy bone



Key concept

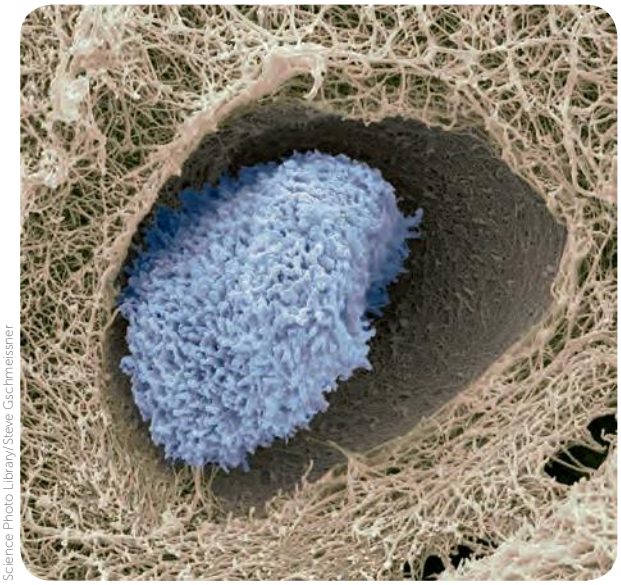
The differing structures of compact and spongy bone enable them to perform different roles in the body.

Structure of cartilage

Like bone, **cartilage** is a connective tissue. It contains numerous fibres made of a protein called collagen. These protein fibres are embedded in a firm matrix of a protein–carbohydrate complex called **chondrin**. This firm matrix enables cartilage to function as a structural support, while the presence of fibres gives cartilage a certain amount of flexibility. Because of these properties, it is found on the surface of bones at the joints, in the trachea and bronchi, and forms the nose, larynx and outer ear.

Cartilage has a firm matrix in which collagen fibres are embedded. Within the matrix are spaces that contain the cartilage cells called **chondroblasts**. These cells produce matrix and gradually become surrounded by it until they are trapped in small spaces called lacunae. Once this has occurred, the cells are considered to be mature and are referred to as **chondrocytes**.

Microscopic structure of cartilage



Science Photo Library/Steve Gschmeissner

FIGURE 8.19
Scanning electron micrograph of cartilage showing a chondrocyte (blue) in a matrix

The collagen fibres in the matrix range in thickness from extremely fine, so that they can just be seen with a microscope, to quite coarse. This variation in the fibrous structure of cartilage is used to classify it into three types (Figure 8.20).

- **Hyaline cartilage** contains many closely packed collagenous fibres throughout the matrix. These fibres are so fine, they are not distinguishable under a light microscope. They give the cartilage strength along with flexibility. Hyaline cartilage makes up the rings of the trachea and bronchi, and is also found in articular cartilage at the ends of bones, where two bones meet to form a movable joint.
- **Elastic cartilage** has conspicuous elastic fibres. It also contains collagenous fibres similar to those in hyaline cartilage, but they are not so closely packed. Elastic cartilage provides flexible elastic support in places such as the external ear. Folding your ear down and letting it go shows how springy this cartilage is.
- **Fibrocartilage** has a coarse appearance from the parallel bundles of thick collagenous fibres that make up this tissue. The fibres are not compacted as much as in hyaline cartilage, and therefore it is able to be compressed slightly. This is ideal for regions where the weight of the body is being supported or where there is a need to withstand heavy pressure. Fibrocartilage

is found in the intervertebral discs of the spinal column, where it provides a cushion between the vertebrae; in the meniscus of the knee joint; and in the tissue joining the two sides of the pelvis.

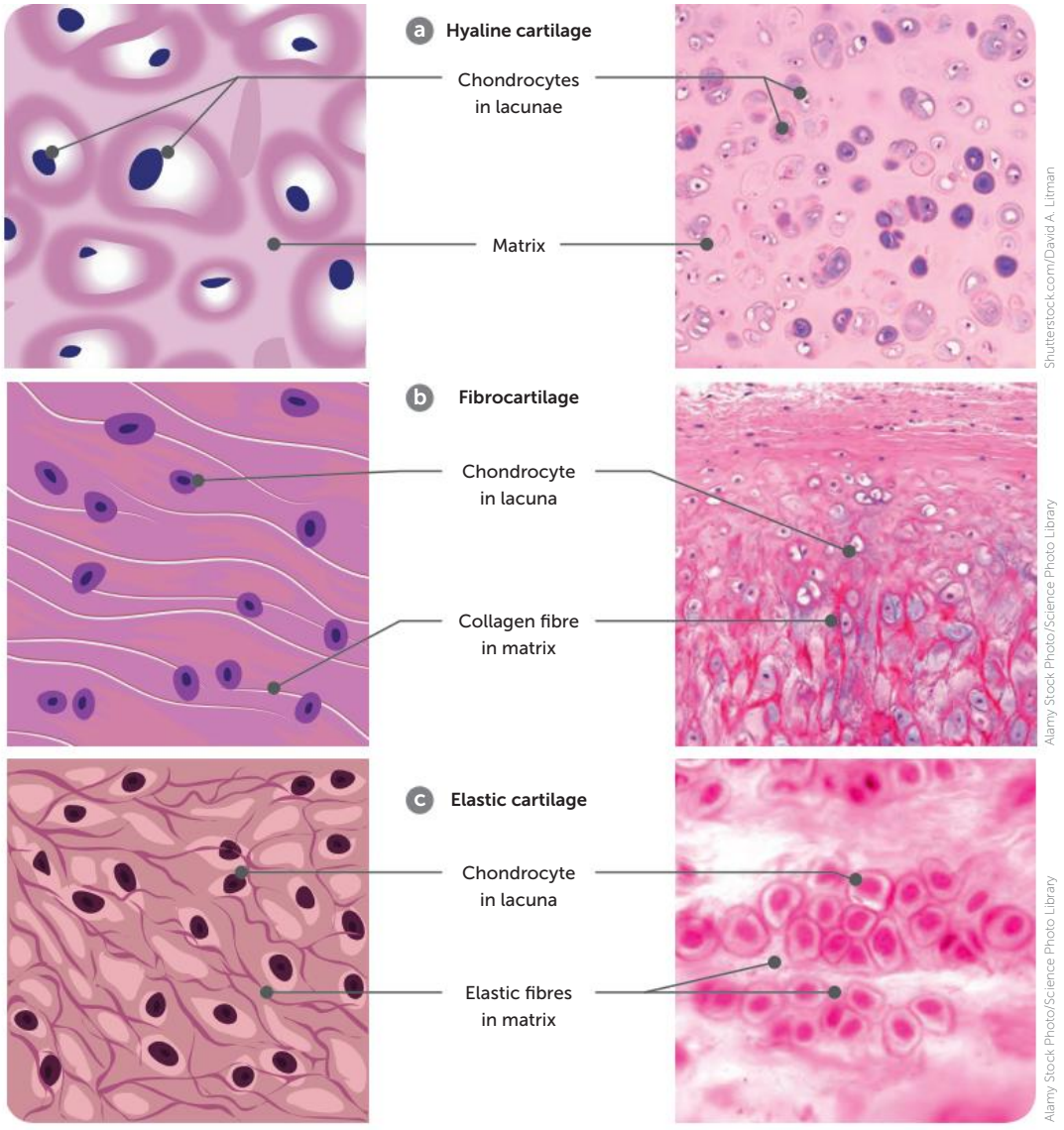


FIGURE 8.20
Structure of the different types of cartilage

Key concept

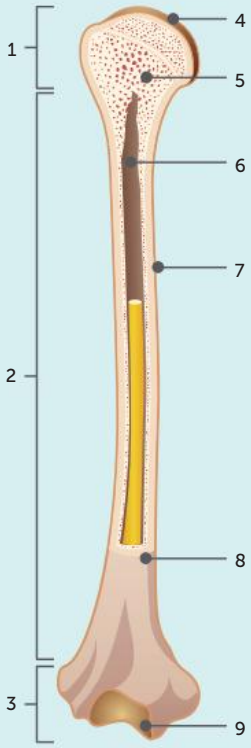
Cartilage may be either hyaline, elastic or fibrocartilage. The variation in the structure of each results in variations in strength, flexibility and elasticity. Therefore, they are found in parts of the body requiring their particular properties.

Cartilage does not contain blood vessels, so all nutrition and waste removal for the cells depends on diffusion through the matrix. This is a slow process and results in the chondrocytes having a slow rate of metabolism and cell division. Injured cartilage therefore takes quite some time to heal. The blood supply to cartilage comes from blood vessels located in the inner layer of the **perichondrium**. This is a fibrous membrane of connective tissue that covers the external surface of cartilage, except where the cartilage forms the articular surface of a joint.

Questions 8.5

RECALL KNOWLEDGE

1 Name the parts of a long bone on the diagram.



2 Describe the structure of an osteon.

3 What type of cartilage is found in the ear?

APPLY KNOWLEDGE

4 Explain the difference between a chondroblast and a chondrocyte.

5 Explain why cartilage takes longer than bone to heal.

8.6 MOVEMENT OF BONES

The site at which two or more bones come together is called a **joint**. Some joints are rather rigid and permit little or no movement. Most, however, allow the bones to move in relation to each other. If the bones fit together tightly, the joint is strong and there is generally very little movement. Loosely fitted joints, on the other hand, allow a great range of movement but are weaker and prone to dislocation.

Joints are frequently classified according to:

- their range of movement (**functional classification**) – e.g. fibrous, cartilaginous or synovial
- the type of connective tissue that binds the bones together (**structural classification**) – e.g. immobile, slightly movable or freely movable.

In the discussion that follows, the joints of the skeleton are described using a combination of both classifications.

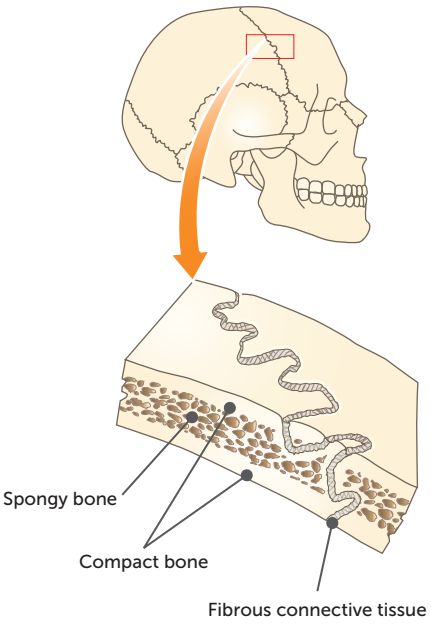


FIGURE 8.21 Sutures of the skull are an example of a fixed or fibrous joint

Types of joints

Fibrous or fixed joints

When no movement occurs between the bones concerned, the joint is described as **fibrous** (or **fixed** or **immovable**). The bones are held in place by fibrous connective tissue, as is the case with the sutures of the skull. It is very difficult to damage this type of joint, as it is so strong that the bone is usually broken, resulting in a fractured skull, before the joint gives way. This joint also occurs between the teeth and the jaw.

Cartilaginous or slightly movable joints

Cartilaginous joints are held in place by cartilage, which allows slight movement to occur. The junction of the two pelvic bones (the pubic symphysis), joints between adjacent vertebrae, and the joints between the ribs and the sternum are examples of slightly movable or cartilaginous joints.

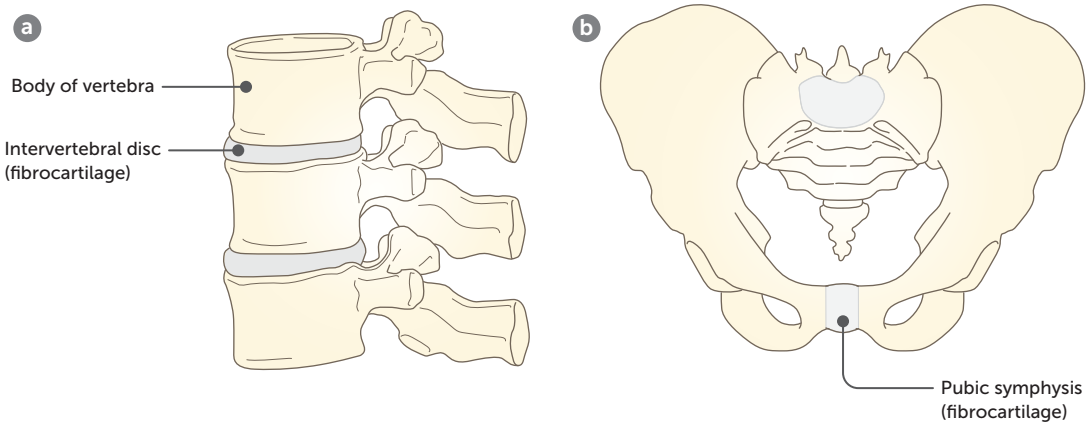


FIGURE 8.22 Slightly movable joints occur between **a** the vertebrae, and **b** the pubic bones

Synovial or freely movable joints

Most joints of the body are **freely movable**, with the amount of movement limited by ligaments, muscles, tendons and adjoining bones. These joints, also known as **synovial joints**, occur at the shoulder, elbow, wrist, fingers, hip, knee, ankle and toes. (Their structure is discussed in more detail in the next section.) These are the most commonly injured joints in sporting and other accidents.

Synovial joints are categorised by the type of movement that occurs between the articulating surfaces of the bones.

- **Ball-and-socket joints** form when the spherical head of one bone fits into a cup-like cavity of another. There are two such joints in the human body: the head of the humerus (upper arm bone) fits into a depression in the scapula (shoulder blade); and the head of the femur (thigh bone) articulates with the pelvis.
- **Hinge joints** allow movement in one plane only, much like that of a hinged door. They form when the convex surface of one bone fits into the concave surface of another. The best examples of this type of joint are the elbow and the knee, but it also occurs at the ankle, and between the bones of the fingers and toes.
- **Pivot joints** are formed when the rounded, pointed or conical end of one bone articulates with a ring, formed partly by bone and partly by a ligament. The best example is the joint between the first vertebra, on which the head is balanced (the atlas), and the second vertebra (the axis). This

allows the head to rotate. Another pivot joint is between the radius and ulna of the forearm; here the joint allows rotation of the hand.

- **Gliding joints** allow movement in any direction in a side-to-side or back-and-forth motion, restricted only by the ligaments or bony processes surrounding the joint. Gliding joints are found between carpal bones, tarsal bones, the sternum and clavicle, and the scapula and clavicle.
- The only true **saddle joint** in the body is where the thumb joins the palm of the hand. The two bones forming the joint are both saddle-shaped – that is, concave in one direction and convex in the other. They fit together in such a way that they allow both side-to-side and back-and-forth movements.
- **Condyloid** (or **ellipsoid**) **joints** have one surface of bone slightly convex that fits into a slightly concave depression in another bone. Such joints occur between the radius and the carpal bones, the metacarpal bones and the phalanges of the fingers, and between the metatarsal bones and the phalanges of the toes. They allow movement in two directions, such as up and down and side to side. For example, hold your index finger erect. You can move it up and down as if beckoning someone, but you can also move it from side to side.

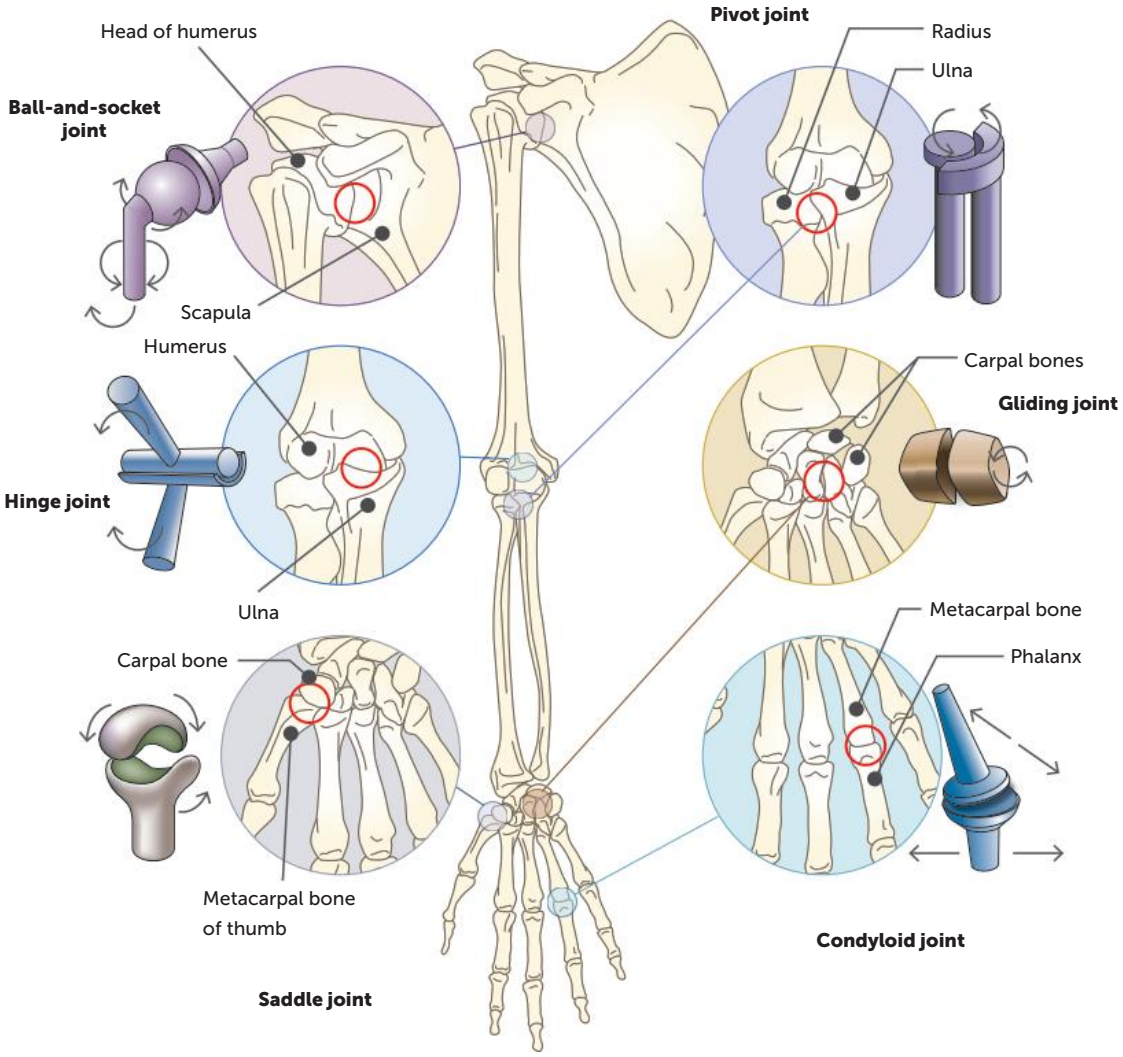


FIGURE 8.23 Types of synovial joints

Types of joints
This website provides more information about the different types of joints.

Movement at joints
Test your knowledge of the bones and joints of the body.

Structure of a synovial joint

Freely movable joints are also called synovial joints because there is a space, or **synovial cavity**, between the articulating surfaces of the bones. A synovial membrane surrounds the synovial cavity, and there is articular cartilage on the bone surfaces.

9780170449090

The knee joint, where the femur meets the tibia, is a typical synovial joint. The patella, or kneecap, is also part of the structure of this joint. Figure 8.24 shows the following parts of the joint.

- An **articular capsule** surrounds and encloses the joint. The capsule comprises two layers.
  - The **fibrous capsule** is the outer layer, consisting of dense, fibrous connective tissue attached to the periosteum of the articulating bones. Its flexibility permits movement at the joint, whereas its strength resists dislocation. The fibrous capsule is one of the principal structures that hold the bones together.
  - The **synovial membrane** is the inner layer of the capsule. It consists of loose connective tissue, the inner surface of which is well supplied with blood capillaries. The synovial membrane lines the entire joint cavity, except the articular cartilages and a structure called the articular disc (if present).
- **Synovial fluid**, secreted by the synovial membrane, fills the synovial cavity and forms a thin film over surfaces within the capsule. It lubricates the joint, helps to keep the articulating surfaces from making contact with one another, provides nourishment for the cells of the articular cartilage, and contains phagocytic cells that remove micro-organisms and any debris resulting from wear and tear at the joint. The synovial fluid is similar in appearance and consistency to egg white. Only a small amount of fluid is normally present (about 0.5 mL in the case of the knee joint). However, this amount may increase, especially in a joint that is injured or inflamed. In such cases, enough fluid may be produced to cause swelling and discomfort.
- **Articular cartilage** covers the articulating surfaces of the bones forming the joint. This tissue provides a smooth surface for movement.
- **Articular discs** occur in some synovial joints. In the knee, there are **menisci** (singular: **meniscus**) consisting of fibrocartilage extending inward from the articular capsule. They divide the synovial cavity into two, meaning that synovial fluid can be directed to the areas of greatest friction. A tearing of the meniscus, commonly referred to as torn cartilage, often occurs in athletes.
- **Bursae**, or little sacs of synovial fluid, are another feature of some joints. These are positioned so that they prevent friction between:
  - a bone and a ligament or tendon
  - a bone and the skin where a bone inside a joint capsule is near the body surface.
- **Ligaments** hold the bones together in many joints (e.g. the ligaments of the knee joint shown in Figure 8.24).

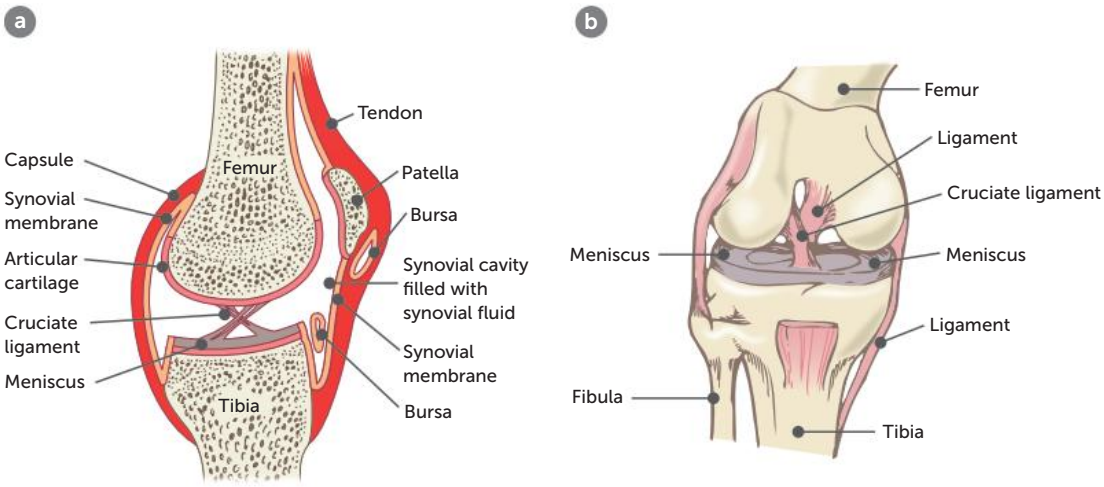


FIGURE 8.24 The knee joint: **a** in section, viewed from the side; and **b** viewed from the front to show ligaments

Key concept

The structure of synovial joints, including the synovial capsule, synovial fluid and articular cartilage, allows the joints to move freely while maintaining their strength.

Keeping joints together

Several factors keep the articular surfaces of synovial joints together. First, there is the fit of the articulating bones – for example, the way the head of the humerus fits into the socket of the scapula to form the shoulder joint (see Figure 8.23). Second, there is the strength of the joint ligaments holding the bones together. (The hip joint illustrates this well.) A third factor is the tension provided by the muscles around the joint: in the knee joint, the fibrous capsule is formed principally from tendons attached to the muscles acting on the joint.

Movement at a joint

Each joint is capable of specific types of movements.

Flexion and extension

Flexion, or bending, decreases the angle between the articulating bones. This means that the bones come closer together. For example, when the elbow is flexed, the lower arm (with the radius and ulna) moves closer to the upper arm (with the humerus).

Extension, or straightening, increases the angle between the articulating bones, moving the bones further apart. For example, when the knee is extended, the lower leg (with the tibia and fibula) moves further away from the upper leg (with the femur).

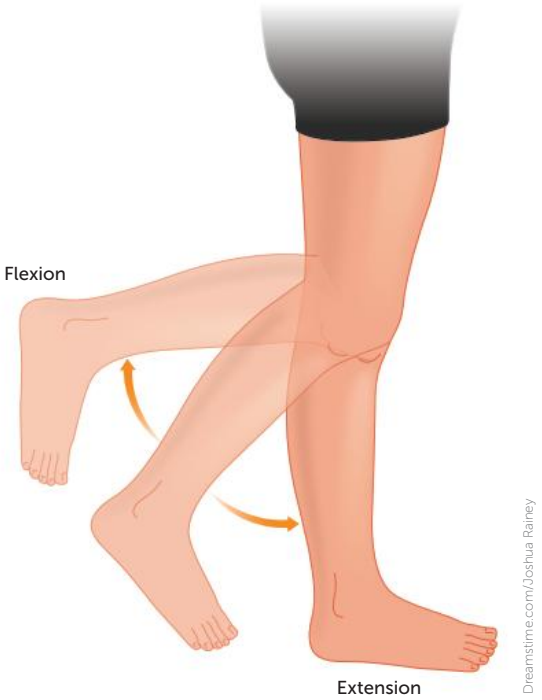


FIGURE 8.25 Flexion and extension of the knee joint

Abduction and adduction

Abduction is movement away from the midline of the body. For example, lifting the arms upwards and away from the body is abduction; while movement towards the midline of the body is **adduction** (e.g. when returning the arms to the sides after abduction).

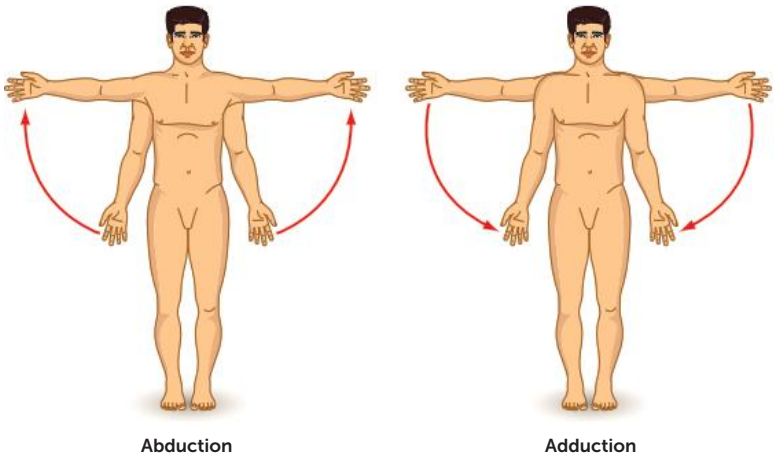


FIGURE 8.26 Abduction and adduction of the arm

Rotation

Rotation is the movement of a bone around its long axis – for example, turning the head from left to right occurs due to rotation at the joint between the first two vertebrae.

FIGURE 8.27
Rotation of the first two vertebrae allows the head to be turned

Questions 8.6

RECALL KNOWLEDGE

1. List the types of joints according to their structural classification.
2. List two examples of fibrous joints.
3. Define 'synovial joints'.
4. Describe the type of joint present in the elbow.
5. Describe the function of synovial fluid.
6. State the type of movement that occurs when kicking a football.

APPLY KNOWLEDGE

7. Describe the difference between a tendon and a ligament.
8. Explain why the shoulder joint has a large range of motion.
9. A joint affected by arthritis often has damage to the articular cartilage. These joints are often swollen. Suggest the reason for the swelling.

8.7 EFFECTS OF AGEING ON THE MUSCULOSKELETAL SYSTEM

Bone is living tissue. A person's bone mass continues to grow until about the age of 30 years. After this, the bones gradually begin to deteriorate. There is a gradual decrease in bone density as the bones lose calcium and other minerals. This is particularly pronounced in women after menopause.

Osteoporosis

If the loss of bone mass becomes sufficient to impair normal functioning, it is called **osteoporosis**. As bone density decreases, the risk of fractures increases so that even minor bumps or falls can result in serious fractures. A fracture is often the first sign that a person has osteoporosis.

The bones most likely to be affected by osteoporosis are the vertebrae, ribs, pelvis, wrist and upper arm, although any bone can be affected.

To prevent osteoporosis, people need an adequate calcium intake in their diet, an adequate amount of vitamin D (either through exposure to sunlight or by dietary intake), and plenty of exercise.

Treatment for osteoporosis includes lifestyle changes to increase calcium intake, vitamin D production and exercise. Smoking contributes to the development of osteoporosis, so quitting smoking is an important lifestyle change for smokers. For some patients, medication can be used to prevent or to treat the condition.

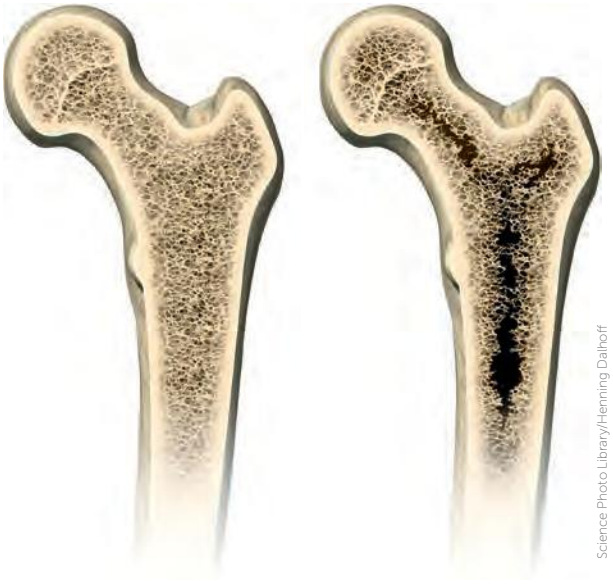


FIGURE 8.28
Cross-section of a normal bone and a bone with osteoporosis

Osteoarthritis

Osteoarthritis is a gradual change in the joints that occurs over time and is frequently associated with ageing. However, other factors, including irritation of the joints, wear and abrasion, may also be involved. In this disease, the joint cartilage deteriorates, and so the bone surfaces are no longer protected. The exposed bone begins to wear away and bony spurs or growths may develop from the exposed ends of the bone forming the joint. These growths and spurs decrease the space within the joint cavity, restricting movement of the joint.

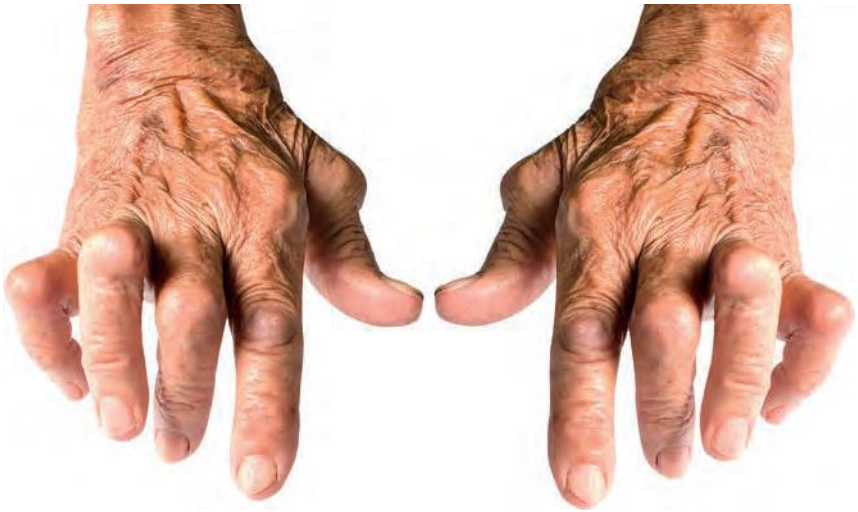


FIGURE 8.29
The bony spurs that develop in osteoarthritis can cause deformities of the hands, feet and limbs, and swelling of the joints

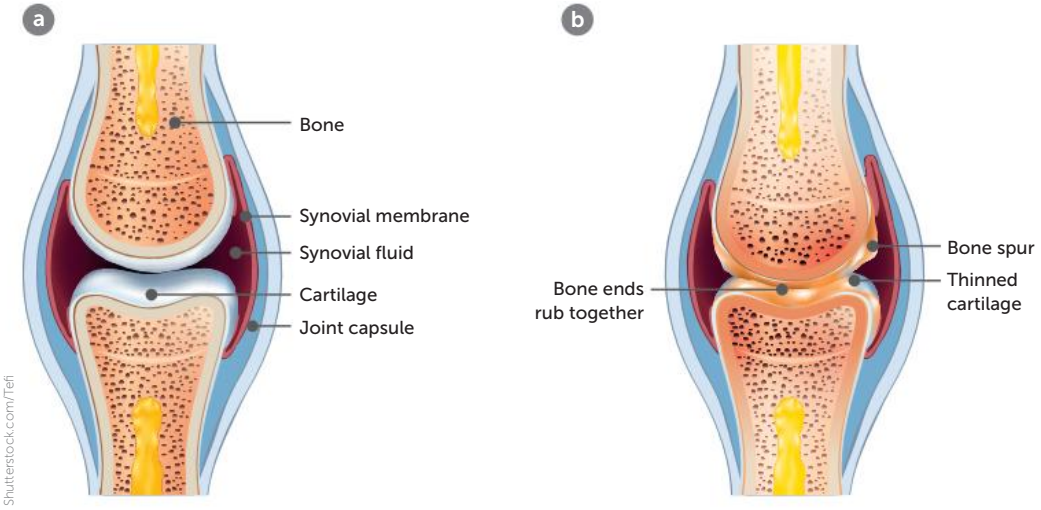


FIGURE 8.30
a Normal joint; **b** Joint affected by osteoarthritis

WS
8.3 Joints

The symptoms of osteoarthritis often appear in middle age, and almost everyone has some symptoms by the age of 70. However, these symptoms may be minor. Before the age of 55, osteoarthritis occurs equally in men and women. After this age, it is more common in women. The most common symptoms are pain and stiffness in the joints; the pain is often more severe after exercise and when weight or pressure is put on the joint. Those who suffer from osteoarthritis notice a rubbing, grating or crackling sound when they move the joint.

There is no known cure for osteoarthritis. Treatment may include medication to relieve pain, physiotherapy to strengthen muscles around the affected joints, surgery to realign bones or joint replacement surgery.

Key concept

Osteoarthritis and osteoporosis are diseases affecting the skeletal system that are more prevalent with increasing age.

Questions 8.7

RECALL KNOWLEDGE

1. Define 'osteoporosis' and 'osteoarthritis'.
2. State what can be done to prevent osteoporosis.
3. By what age is it common to experience some osteoarthritis?

APPLY KNOWLEDGE

4. Explain why osteoporosis often leads to bone fractures.
5. Explain why osteoarthritis causes painful joints.

9780170449090

CHAPTER 8 ACTIVITIES

ACTIVITY 8.1 Investigating fast- and slow-twitch fibres

Our skeletal muscles are composed of two types of fibres, described as slow-twitch and fast-twitch fibres. As their name suggests, slow-twitch fibres are those that contract gradually. In doing so, they produce little power but are resistant to fatigue. This makes slow-twitch fibres ideal for situations where endurance is important. Fast-twitch fibres, on the other hand, are suitable for short bursts of activity. They contract more quickly and in doing so they produce more power.

What to do

The aim of this activity is to determine the muscle fibre composition of one of the muscles in your thigh – the quadriceps muscle. To do this, you will have to do a wall sit. A wall sit is when you stand with your back against a wall, then slowly slide down the wall into a seated position so that your thighs are parallel to the ground. Your back should still be against the wall. In this position, your muscle fibres will be trying to shorten but will not be able to do so completely. These muscle contractions will enable you to roughly determine the proportions of slow-twitch and fast-twitch fibres you have in your upper legs.

Work in pairs, with one person acting as subject and the other as timer before swapping roles.

1. Move to a clear section of wall. Lean against the wall and then assume the wall sit position with thighs parallel to the floor.
2. Use a stopwatch to time how long you are able to hold the position; stop when you can no longer tolerate the burning sensation in your upper legs. Do not hold the position for more than two minutes.
3. Record the time you were able to maintain the wall sit position.
4. Change roles and time your partner doing the wall sit.
5. Compare your time with the times in the table below and determine the most likely composition of the fibres in your upper leg muscles.
6. Draw up a suitable table and record the results for all class members.

TIME OF WALL SIT	TYPE OF MUSCLE FIBRE
Less than 30 seconds	It is likely that you have more fast-twitch than slow-twitch fibres in your upper leg muscles.
More than 30 seconds, but less than a minute	It is likely that around 50% of the fibres in your upper leg muscles are slow twitch. The closer you approach to one minute, the nearer the number of slow-twitch fibres is to 50%.
More than a minute	It is likely that your upper leg muscles consist mainly of slow-twitch fibres.

Studying your results

1. Describe what was happening to your muscle fibres while you were in the wall sit position.
2. List the types of activities that would use fast-twitch fibres and those that would use slow-twitch fibres.
3. Study the class data.
   - **a** Does it appear that students who participate in sports or activities that require endurance are better able to hold the wall sit position?
   - **b** Does it appear that students who participate in sports or activities that require short bursts of energy did not last as long in this activity?

Summary

Write a short paragraph outlining the difference between slow-twitch and fast-twitch muscle fibres and the activities for which each would be suited.

ACTIVITY 8.2 Studying a long bone

In this activity, you will study the structure of a long bone to see how well it is suited to its functions of support and movement when pulled by the muscles attached to it.

You will need

An uncooked long bone (from a cow or a sheep); a long bone cut lengthwise (in longitudinal section); a hand lens or magi-lamp

What to do

1. Examine the bone carefully and observe the smooth, bluish-white coverings on the ends. This is the articular cartilage.
2. Around the bone is a sheath of fibrous tissue, the periosteum. Locate any blood vessels in the periosteum and any places where muscles may have been attached. Muscles will appear as red meat attached to the periosteum.
3. Examine the longitudinal section of bone with a lens and identify the marrow cavity, the spongy or cancellous bone, and the compact bone.

Studying your observations

1. Draw a diagram of a longitudinal section of long bone. Label the following structures on your diagram: periosteum, articular cartilage, epiphysis, diaphysis, compact bone, spongy bone and marrow cavity.
2. Describe the function of the articular cartilage. How do the location and texture of the cartilage relate to its function?
3. What is the purpose of the blood vessels in the periosteum? Why does bone require a blood supply?
4. Describe how muscles are attached to the bone.
5. What is the thickness of the compact bone (in millimetres) at the epiphyses and the diaphysis?
6. Explain the differences in location and structure of the spongy bone and the compact bone. Relate these differences to the functions of the two types of bone.
7. Why aren't long bones solid? What is the purpose of the marrow cavity? Describe the marrow that fills the marrow cavity.
8. Why do the ends of the bone, the epiphyses, have a greater diameter than the shaft of the bone, the diaphysis? Suggest at least two reasons.

ACTIVITY 8.3 Investigating the composition of bone

As bone forms, inorganic salts are deposited in the matrix between the cells. In this activity, you will investigate the effect of removing some of these inorganic salts.

You will need

Two small bones or pieces of bone; two 100 mL beakers; forceps; 2 mol/L nitric acid

What to do

1. Place a small bone, or piece of bone, in a beaker and cover it with nitric acid. In the second beaker, place a similar piece of bone and cover it with water.

9780170449090

2 Leave the bones to stand for at least two days.
3 Remove the bones and rinse them under running water.
4 Feel, and try bending, each of the two bones.

Studying your observations

1 Describe any differences that you observed between the bone left in acid and the bone left in water.
2 Propose a hypothesis to account for any differences that you observed.
3 If a person does not consume enough calcium in their diet for normal body functioning, calcium is removed from the bones. What would be one of the symptoms of severe dietary calcium deficiency?

9780170449090

CHAPTER 8 SUMMARY

- The muscular and skeletal systems work together to support and move the body.
- Muscle cells can contract and shorten.
- Skeletal muscles move the bones and can contract under conscious control.
- Smooth muscle in the internal organs is involuntary muscle and is not under conscious control.
- Cardiac muscle makes the heart contract, pushing blood around the body.
- Muscles have extensibility and elasticity; they can be stretched and then contract.
- Skeletal muscle cells are held in bundles by connective tissue.
- Each skeletal cell is called a muscle fibre; the plasma membrane is called the sarcolemma and the cytoplasm is called the sarcoplasm.
- Parallel myofibrils in the sarcoplasm are composed of thick myofilaments made of myosin and thin myofilaments made of actin.
- The overlap of the myofilaments causes striations that divide the myofibril into sarcomeres.
- The sliding filament theory describes how muscles contract. As the thin filaments slide over the thick filaments, the sarcomere shortens.
- Energy, from ATP, is used during muscle contraction.
- Muscles are attached to bones by tendons so that the joint bends when the muscle contracts.
- Antagonists are pairs of muscles. The agonist is the muscle doing the action.
- Synergists help the agonist by producing the same movement or steadying the joint.
- Muscle tone is achieved by partial contraction of skeletal muscles due to different fibres contracting at one time. This allows a person to maintain their posture.
- The skeletal system supports the body, facilitates movement, protects internal organs, produces blood cells, and stores and releases minerals and fat.
- Bones may be classified as flat, irregular, long, sesamoid or short.
- The skeleton can be divided into the axial skeleton (the central bones) and the appendicular skeleton (the limbs, shoulder and pelvis).
- Long bones contain a shaft called the diaphysis, which is made of compact bone and contains a cavity with yellow bone marrow, and ends called epiphyses, made of spongy bone with red marrow.
- Compact bone is made up of many osteons or Haversian systems.
- Each osteon has a central canal surrounded by concentric lamellae. Osteocytes are located in small spaces between the lamellae called lacunae. Canals called canaliculi join the lacunae.
- Spongy bone is composed of irregular bony plates called trabeculae. The osteocytes are located between the trabeculae.
- Cartilage is made up of protein fibres called collagen that are embedded in a matrix called chondrin. The chondroblasts are located in the matrix.
- The arrangement of collagen fibres determines whether the cartilage is hyaline, elastic or fibrocartilage.
- A joint occurs when two or more bones come together.
- Types of joints are fibrous, cartilaginous, freely movable, ball and socket, pivot, gliding, saddle or condyloid, based on their range of movement and the connective tissue joining them.
- Freely movable joints are called synovial joints due to the synovial cavity. The joint is enclosed by the articular capsule and contains synovial fluid.
- Ligaments connect two bones.
- Joints move in flexion, extension, abduction, adduction and rotation.
- Osteoporosis and osteoarthritis are more likely with increased age.

9780170449090

CHAPTER 8 GLOSSARY

Abduction Movement of a limb away from the midline of the body

Actin One of the contractile proteins of skeletal muscle; makes up the thin myofilaments of a myofibril

Adduction Movement of a limb towards the midline of the body

Agonist Muscle that causes the desired action; often referred to as the prime mover

Antagonist Muscle that has an action opposite to that of the prime mover (*see* agonist)

Appendicular skeleton The part of the skeleton made up of the bones of the upper and lower limbs, and the bones of the shoulders and pelvis

Articular capsule The sac-like envelope in the cavity of a synovial joint; also called joint capsule

Articular cartilage Cartilage that covers the surfaces of bones at a joint

Articular disc An alternative name for meniscus

Articulation *see* joint

Axial skeleton The part of the skeleton that forms the central axis of the body; consists of skull, vertebral column, ribs and sternum

Ball-and-socket joint Joint consisting of a spherical bone fitting into a cup-like cavity of another bone

Belly The fleshy portion in the middle of a muscle

Biceps The muscle on the front part of the upper arm

Bursa A small sac or cavity filled with fluid; found at friction points of a movable joint; plural: bursae

Canaliculi Small canals in the matrix of bone; join the spaces that contain bone cells; singular: canaliculus

Cancellous bone Bone that contains many large spaces; appears 'spongy'

Cardiac muscle The muscle that forms the wall of the heart

Cartilage A type of connective tissue containing fibres of collagen

Cartilaginous joint A joint at which only limited movement occurs between the bones; the bones are held in place by cartilage as between the ribs and sternum; also called a slightly movable joint

Central canal A hollow that runs through the centre of the spinal cord; also the central channel in an osteon of compact bone (may be called the Haversian canal)

Chondrin A matrix of protein and carbohydrate in cartilage

Chondroblast A cell that forms the fibres and matrix of cartilage

Chondrocyte A mature cartilage cell

Compact bone A dense form of bone

Condyloid joint A synovial joint in which one surface of bone is slightly convex and fits into a slightly concave depression in another bone; also known as an elipsoid joint

Connective tissue Tissue providing support for body organs

Contraction Shortening of a muscle

Diaphysis The shaft of a long bone

Elastic cartilage Cartilage that contains elastic fibres

Elasticity The ability of muscle fibres to return to their original length after being stretched

Ellipsoid joint *see* condyloid joint

Epiphysis The end of a long bone; plural: epiphyses

Extensibility The ability of muscle fibres to be stretched when pulled

Extension Lengthening of a muscle

Fibrocartilage A type of cartilage that contains bundles of fibres

Fibrous capsule The external layer of an articular capsule

Fibrous joint A joint at which no movement occurs between the bones; the bones are held in place by fibrous tissue, as between

9780170449090

the bones of the skull; often called a fixed or immovable joint

Fixator A muscle that contracts to immobilise a joint

Fixed joint *see* fibrous joint

Flexion To bend; a movement that decreases the angle between articulating bones

Freely movable joint *see* synovial joint

Functional classification Classification of a joint based on its range of motion

Gliding joint A joint that allows movement in any direction

Haversian canal A network of tubes in compact bone occupied by nerves and blood vessels

Haversian system *see* osteon

Hinge joint A joint that allows movement in one plane only

Hyaline cartilage Flexible supporting cartilage

Immovable joint *see* fibrous joint

Insertion The end of a muscle fixed to the movable bone

Involuntary muscle Muscle that is not under conscious control; found in walls of internal organs; also called non-striated muscle, smooth muscle or plain muscle

Joint The connection between two bones

Lacunae Space in the matrix of bone or cartilage occupied by a cell; singular: lacuna

Lamellae Concentric rings that make up compact bone; singular: lamella

Ligament Fibrous tissue that attaches one bone to another bone

Matrix Non-cellular material between the cells of a tissue

Meniscus A cartilaginous disc found in the knee joint; divides the cavity into two parts; plural: menisci

Muscle fibre The long cylindrical cells that make up skeletal muscles

Muscle tone Partial contraction of skeletal muscles

Myofibril A thread-like structure found in the cytoplasm of muscle fibres

Myofilament One of the structures that make up the myofibrils of skeletal muscle fibres

Myosin One of the contractile proteins of skeletal muscle; makes up the thick myofilament of a myofibril

Origin The end of a muscle that is fixed to the stationary bone

Osteoarthritis Deterioration of joint cartilage due to age or injury, to the point where the bone surfaces are not protected

Osteocyte A mature bone cell

Osteon The basic unit of structure of compact bone; consists of a central canal surrounded by concentric layers of hard matrix and bone cells; also called the Haversian system

Osteoporosis Reduced bone density due to ageing, resulting in increased risk of fractures

Perichondrium A membrane that covers some types of cartilage

Periosteum The dense, fibrous outer covering of a bone

Pivot joint A joint that allows rotation, such as between the radius and ulna

Posture The way a person holds their body when standing or sitting

Prime mover An alternative name for agonist

Red bone marrow A region of some bones where blood cell production takes place

Rotation Movement of a bone around its long axis

Saddle joint The joint where the thumb is attached to the palm

Sarcolemma A thin, transparent membrane surrounding a muscle cell

Sarcomere The contractile unit of skeletal muscle; consists of actin and myosin filaments

Sarcoplasm The cytoplasm of striated muscle fibres

Skeletal muscle Muscle attached to bones, under voluntary control; also called voluntary or striated muscle

9780170449090

Sliding filament theory The theory used to explain muscle contraction

Smooth muscle *see* involuntary muscle

Spongy bone An alternative name for cancellous bone

Structural classification Classification of a joint based on the type of connective tissue holding the joint together

Synergist A muscle that acts indirectly in steadying a joint during a particular movement

Synovial cavity The space between articulating bones in a synovial joint

Synovial fluid Fluid that fills the cavity of a synovial joint

Synovial joint A freely movable joint; the amount of movement possible is limited by ligaments, muscles, tendons and adjoining bones

Synovial membrane The inner layer of a capsule around a synovial joint

Tendon Fibrous tissue that attaches muscle to bone

Trabeculae Bony plates that criss-cross with many others to make up spongy bone; singular: trabecula

Triceps The muscle at the back of the upper arm

Yellow bone marrow The region of a bone where fat is stored

9780170449090

CHAPTER 8 REVIEW QUESTIONS

Recall

1 List the three types of muscle, and state one location of each type.
2 Give a definition and example for: agonist, synergist, ligament.
3 Briefly describe five main functions of the skeletal system.
4 Describe the external and internal structure of a typical long bone.
5 Draw and label a diagram of the microscopic structure of compact bone.
6 State the function of the following microscopic structures of compact bone: central canal; lamellae; lacunae; osteocytes; canaliculi.
7 Describe the factors that limit the amount of movement about a joint.
8 Distinguish between the three main types of joints based on:
   a structure
   b function.
9 List the functions of synovial fluid.
10 Draw a labelled diagram of a synovial joint.

Explain

11 Explain how the sliding filament theory accounts for the action of muscles.
12 Explain how muscle tone contributes to a person's posture.
13 Explain how muscles produce movement about a joint. In your answer, distinguish between the roles of agonists and antagonists.
14 Explain what is meant by the term 'joint', with regard to the skeleton.
15 Differentiate between the axial skeleton and the appendicular skeleton.
16 Describe all the factors that contribute to keeping the articulating surfaces of a synovial joint in contact with each other.
17 Explain why articular discs are important in some joints.

Apply

18 How does muscle tissue differ from the other tissues of the body?
19 Describe the characteristics that would allow you to classify a muscle as skeletal or smooth when viewed on a microscope slide.
20 Compare and contrast the muscles that move the lower arm with those that move the lower leg.
21 Using an example, describe how a fixator muscle can act as a stabiliser to allow other muscles to perform a particular movement.
22 Differentiate between:
   a compact and cancellous bone
   b yellow bone marrow and red bone marrow
   c diaphysis and epiphysis
   d osteon and trabeculae.
23 Use an example to differentiate between:
   a flexion and extension
   b abduction and adduction.
24 Compare and contrast osteoporosis and osteoarthritis.
25 Suggest reasons why epiphyses of long bones are composed of cancellous bone, while the diaphysis is composed of compact bone.
26 Eight types of joints are described in this chapter. Rank them from the type that allows the greatest degree of movement to the one that allows the least.

9780170449090

Extend

27 Use your understanding of the mechanism of breathing and the structure of joints to explain why the ribs are attached to the sternum with cartilage.

28 Explain how the muscular and skeletal systems must work interdependently.

29 In doing the wall sit in Activity 8.1, your quadriceps muscle was contracting isometrically. Muscles are also able to contract isotonically. Find out the difference between these two types of muscle contraction. Then, using the biceps muscle as an example, describe situations that would result in each of these two types of contraction.

30 Sarcopenia is the degenerative loss of skeletal muscle mass and strength associated with ageing. Conduct research to ascertain the effect that ageing has on:

- **a** the number and size of muscle fibres
- **b** changes in muscle contractile properties and the effectiveness of the motor unit
- **c** the effectiveness of the mitochondrial proteins
- **d** how exercise can help a person avoid some of the detrimental effects of ageing.

31 The sliding filament model is more complex than the explanation given above. Find out:

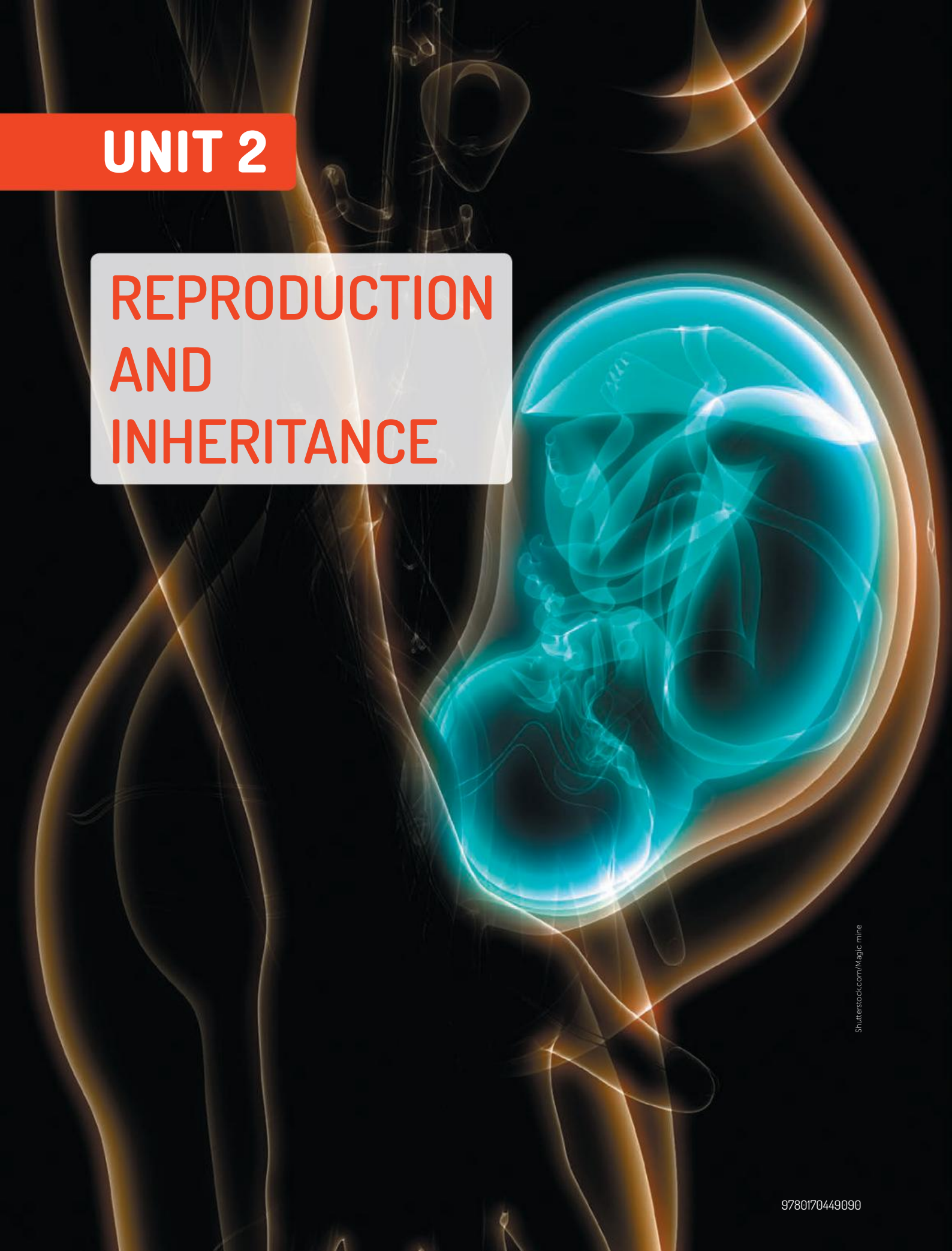
- **a** the shape of the myosin molecules and how they are arranged to make up the thick filaments
- **b** the composition of the thin filaments and the active sites they contain
- **c** the function of the protein tropomyosin
- **d** the location and role of the protein titin.

32 Conduct research to find out how smooth and cardiac muscles contract.

9780170449090

UNIT 2

REPRODUCTION AND INHERITANCE



Shutterstock.com/Magic mine

9780170449090

9

DNA DETERMINES THE STRUCTURE AND FUNCTION OF CELLS

UNIT 2 CONTENT

SCIENCE INQUIRY SKILLS

» select, construct and use appropriate representations, including models of DNA replication, transcription and translation, Punnett squares, pedigrees and karyotypes, to communicate conceptual understanding, solve problems and make predictions

SCIENCE AS A HUMAN ENDEAVOUR

» discoveries made through the use of modern biotechnological techniques have increased understanding of DNA and gene expression

SCIENCE UNDERSTANDING

DNA

» DNA occurs bound to proteins in chromosomes in the nucleus and as unbound DNA in the mitochondria

» DNA stores the information for the production of proteins that determines the structure and function of cells

» the structural properties of the helical DNA molecule, including double-stranded, nucleotide composition and weak bonds involved in base pairing between the complementary strands, allow for its replication

» protein synthesis involves the transcription of a gene on DNA into messenger ribonucleic acid (RNA) in the nucleus, and translation into an amino acid sequence at the ribosome with the aid of transfer RNA

» epigenetics is the study of phenotypic expression of genes, which depends on the factors controlling transcription and translation during protein synthesis, the products of other genes, and the environment

Source: School Curriculum and Standards Authority, Government of Western Australia

9780170449090

9.1 DNA, GENES AND CHROMOSOMES

DNA, short for **deoxyribonucleic acid**, is a molecule found in the cells of all organisms – bacteria, single-celled plants and animals, and all complex plants and animals, including humans. It contains the genetic information that determines the structure of the cell and the way it functions.

Most of the DNA molecules are found in the nucleus of each cell, and are therefore called nuclear DNA. However, there is also a small amount of DNA found in the mitochondria. This DNA is called mitochondrial DNA, or mtDNA. While mtDNA makes up less than 1% of the total DNA in humans, it is important for the functioning of the mitochondria and, therefore, the cell.

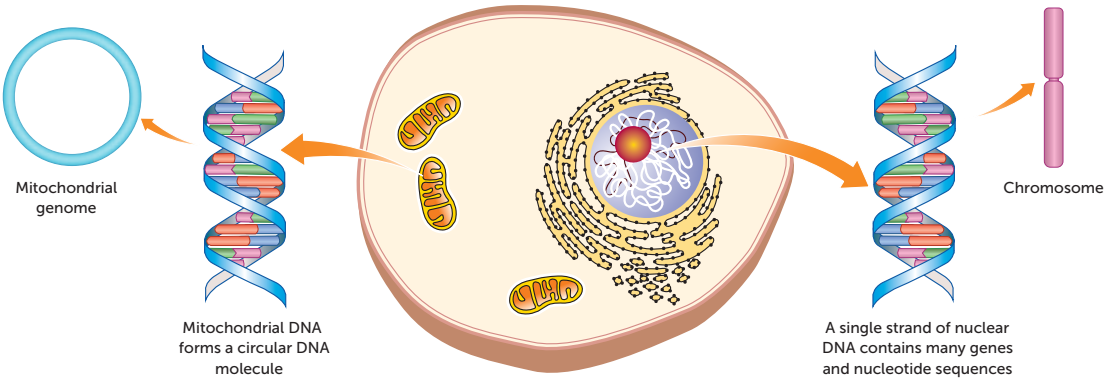


FIGURE 9.1 A cell contains both nuclear and mitochondrial DNA

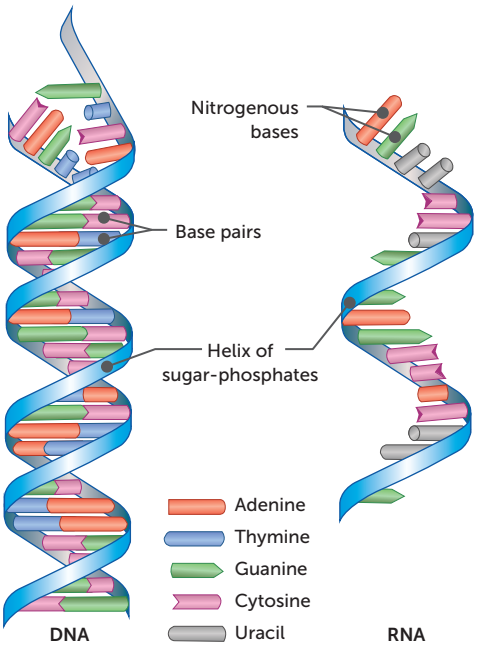


FIGURE 9.2 DNA and RNA are both nucleic acids

Structure of DNA

DNA is one of the two main types of nucleic acids found in the body; the other is RNA, or ribonucleic acid.

DNA is an example of a polymer, a molecule made up of many repeating small units. In the case of nucleic acids, the small repeating units are **nucleotides**. Each nucleotide is composed of a sugar molecule (deoxyribose in DNA), a phosphate group and a nitrogenous base. There are four different nitrogen bases in the DNA molecule: **adenine** (**A**), **thymine** (**T**), **cytosine** (**C**) and **guanine** (**G**).

The sugar molecule of one nucleotide bonds to the phosphate group of another one. This forms a long chain of alternating sugars and phosphates, with side chains of bases. In DNA, two strands join by specific bases being attracted to one another by weak hydrogen bonds. Cytosine on one strand can only bond to guanine on the other strand, while adenine can only bond to thymine. The two strands of DNA twist into a spiral shape called a **double helix**.

9780170449090

The order in which the nitrogenous bases occur in the DNA molecule determines the genetic code. A code of only four letters (A, T, C and G) would not seem to allow many different combinations, but each gene consists of up to 2 million pairs of bases. The number of possible combinations of base pairs is therefore enormous.

Molecules of DNA are in the form of long strands. It is estimated that the length of the DNA molecules in a human cell is between 2 and 3 metres, but the width is only two-millionths of a millimetre. An average human hair is about 40 thousand times thicker! As a comparison, if a DNA molecule were as thick as a pencil (about 8 mm), then the molecule would be about 200 km long. Imagine how difficult it would be to control a pencil 200 km long without breaking it. Yet, the nuclei of human cells have 46 such DNA molecules.

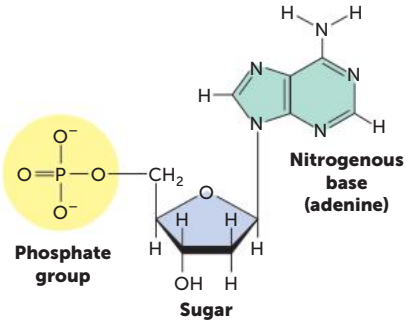


FIGURE 9.3 Structure of a nucleotide

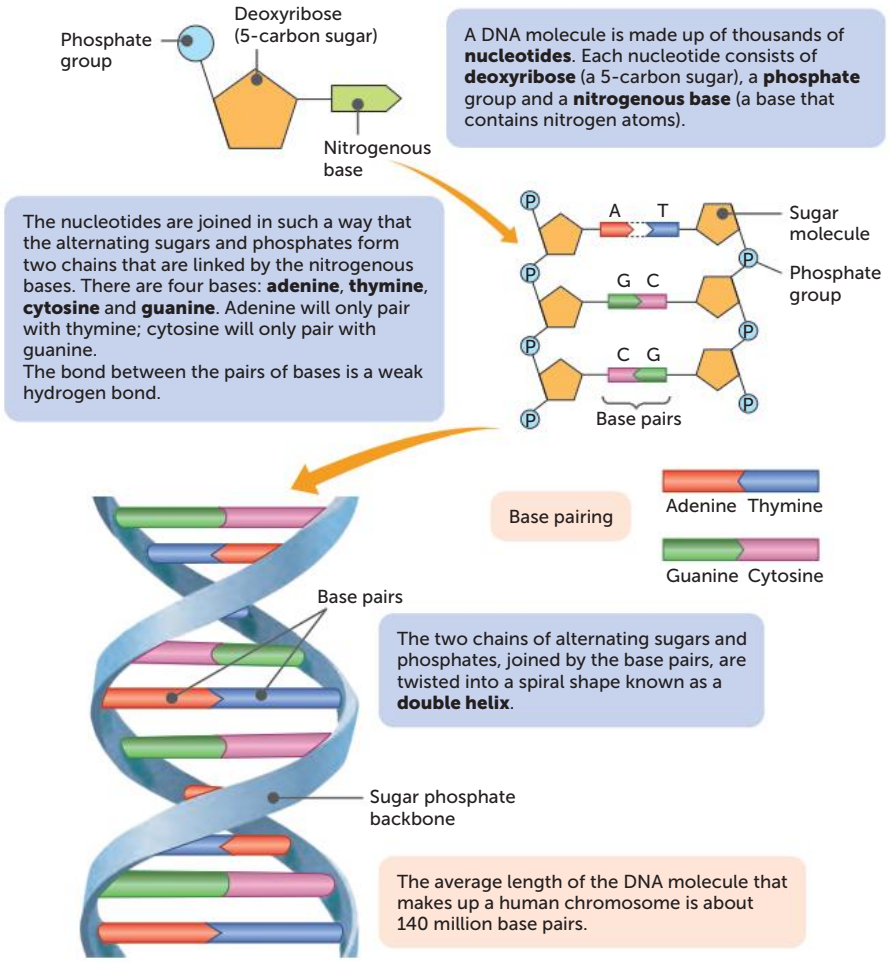


FIGURE 9.4 Structure of a DNA molecule

Key concept

Deoxyribonucleic acid (DNA) is a large molecule made up of many nucleotides composed of sugar, phosphate and nitrogenous bases joined together to form a double-stranded structure coiled into a spiral.

9.1 DNA colouring

DNA from the beginning
Learn more about the experiments that contributed to our knowledge of DNA.

9780170449090

Structure of chromosomes

DNA code
This website shows the discoveries, and the mistakes, made as scientists unravelled the mystery of the DNA code.

How do such long molecules fit inside the cell nucleus? The DNA strands are actually wrapped around a group of eight special proteins called **histones** to form a **nucleosome**. There are many nucleosomes along the length of a DNA molecule.

In a cell that is not dividing, the coiled DNA forms a tangled network called **chromatin**. However, when a cell divides the coiled chromatin becomes even more tightly coiled. These 'super-coiled' structures are large enough to be seen with a light microscope and are called **chromosomes**.

There are 46 chromosomes in a normal human cell. Each chromosome is made up of sections of DNA that code for a particular protein. Each of these sections is called a **gene**.

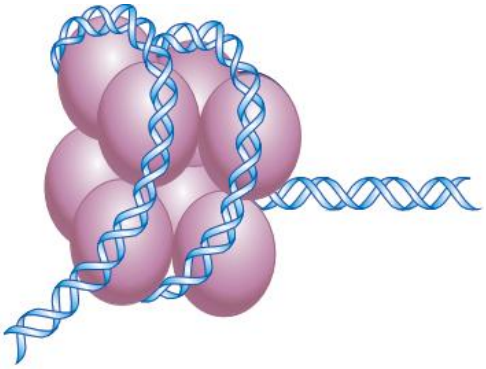


FIGURE 9.5 A nucleosome is made up of DNA wrapped around eight histone proteins

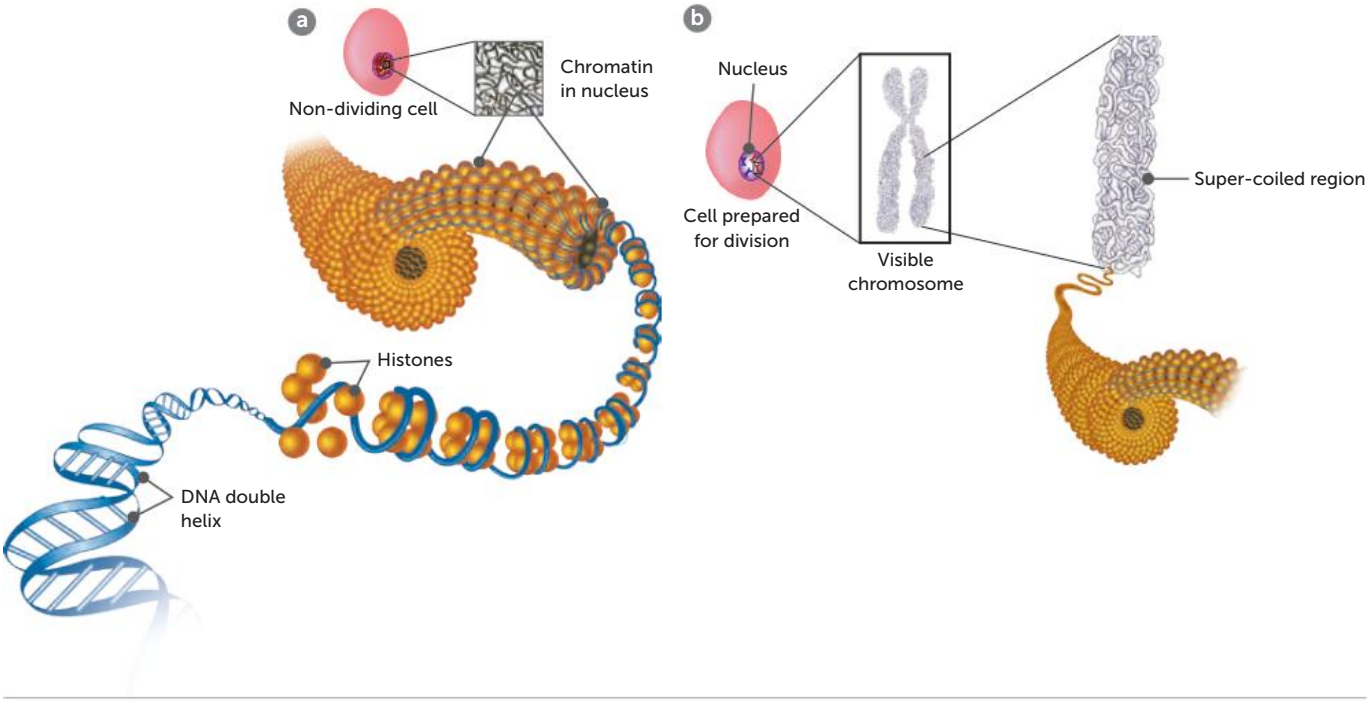


FIGURE 9.6 a When a cell is not dividing, the DNA is wrapped around histones in a tangled network called chromatin; **b** When a cell is dividing, the chromatin becomes super-coiled to form chromosomes

Mitochondrial DNA

Mitochondria are the organelles in the cell where the aerobic phase of respiration occurs, releasing energy for use by the cell. Most of a cell's DNA is located in the nucleus, but a small amount is in the mitochondria. This is called **mitochondrial DNA**, or mtDNA.

9780170449090

There are two important differences between DNA in the nucleus and mtDNA.

- **Nuclear DNA** is in the form of very long strands that are bound to proteins, the histones.
- Mitochondrial DNA is in the form of small circular molecules that are not bound to proteins.

There are about five to ten molecules of mtDNA in each mitochondrion. It has 37 genes, all of which are essential for the mitochondrion to function normally. Twenty-four of the genes contain the code for making transfer RNA (tRNA) molecules, which are involved in protein synthesis. The other 13 genes have instructions for making some of the enzymes necessary for the reactions of cellular respiration.

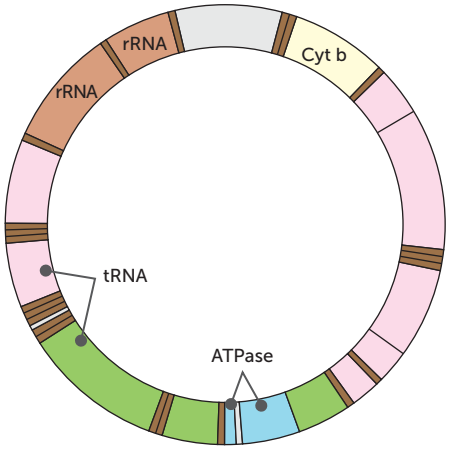


FIGURE 9.7 Model of a molecule of mtDNA showing the location of some of the genes

Key concept

Mitochondrial DNA is a circular molecule not bound to histones. It plays an important role in coding for transfer RNA and enzymes needed for cellular respiration.

Replication of DNA

Cells divide through the processes of mitosis and meiosis. You will learn more about these in Chapter 10. In all cell divisions, the DNA must produce an exact copy of itself. This is known as **DNA replication**.

The first stage of DNA replication is when the two strands of the DNA molecule are separated by the enzyme **helicase**. This separation is possible because the hydrogen bond between the base pairs is weak and is therefore easily broken.

Each strand of the separated section contains half the original information. Each strand serves as a template for the nucleotides that will form the new strand. As the base adenine can only pair with thymine, and cytosine can only pair with guanine, the new strand that forms is identical to the original. Figure 9.8 shows how this process results in the formation of two identical DNA molecules.

Two enzymes play important roles in the synthesis of the new strands. **DNA polymerase** adds the new nucleotides to the new strand, and **DNA ligase** joins short sections of DNA together.

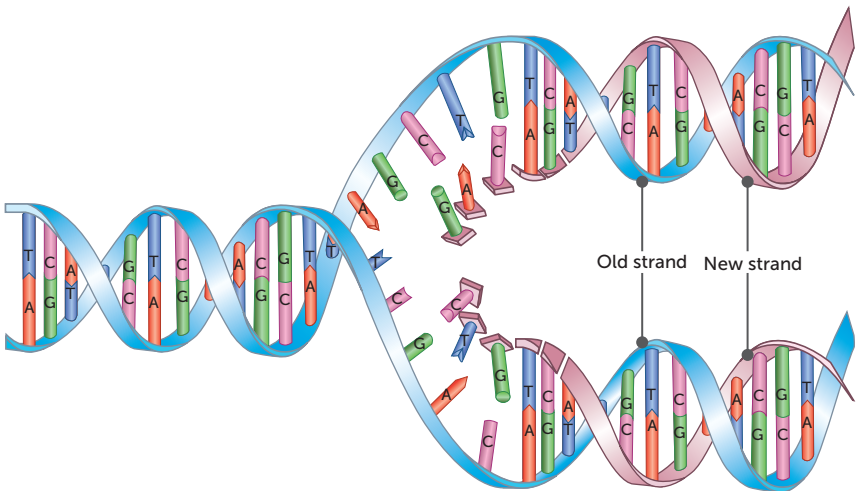


FIGURE 9.8 DNA replication – the two strands separate and act as templates for new strands

9780170449090

Activity 9.1
Modelling DNA structure and replication

Activity 9.2
Extracting DNA

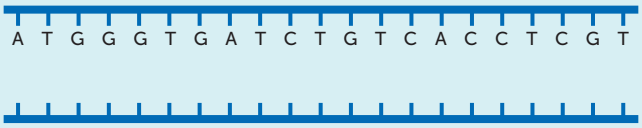
Questions 9.1

RECALL KNOWLEDGE

1. State the location of DNA in the cell.
2. Draw a labelled diagram of a nucleotide.
3. Name the four nitrogenous bases found in DNA.
4. Describe the double helix structure of DNA.
5. Describe the structure of chromatin and chromosomes.
6. Outline the steps of DNA replication.

APPLY KNOWLEDGE

7. State two similarities and two differences between nuclear DNA and mitochondrial DNA.
8. Write the missing bases on the diagram below.



9. Explain the relevance of nucleosomes.

9.2 PROTEIN SYNTHESIS

The genetic code in DNA provides the instructions for protein synthesis – making proteins in the cell. You will recall from Chapter 3 that proteins are produced from amino acids being joined together by peptide bonds. The sequence of bases in DNA controls the order of amino acids, and therefore the type of protein that is produced. Some proteins that you would have heard of are:

- haemoglobin, the oxygen-carrying molecule in red blood cells
- actin and myosin, the proteins involved in muscle contraction
- albumin, which is found in egg white and in human blood plasma
- fibrin, a protein involved in blood clotting
- collagen, the main component of bones, teeth, cartilage, ligaments and tendons
- insulin, a hormone that regulates the concentration of glucose in the blood
- immunoglobulins, which are found in the blood and act as antibodies to combat foreign matter, such as infecting micro-organisms
- amylase, an enzyme that breaks down starch.

Ribonucleic acids

Ribonucleic acid (RNA) is another type of nucleic acid. Like DNA, RNA is composed of a chain of nucleotides. While there are many similarities between DNA and RNA, there are also some key differences.

- The sugar molecule is ribose, not deoxyribose. Ribose has one more oxygen atom than deoxyribose.
- RNA is single stranded, while DNA is double stranded.
- RNA has the bases **cytosine**, **guanine**, **adenine** and **uracil**, while DNA has the bases cytosine, guanine, adenine and thymine. The structures of thymine and uracil are very similar. This means that uracil is also complementary to adenine.
- The RNA strand is able to fold on to itself, forming hydrogen bonds between complementary bases.

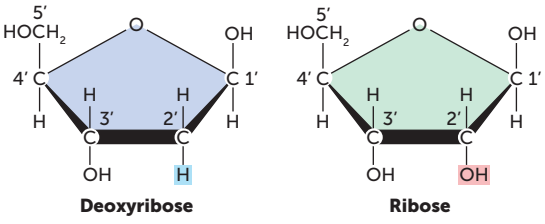


FIGURE 9.9
Differences between deoxyribose and ribose

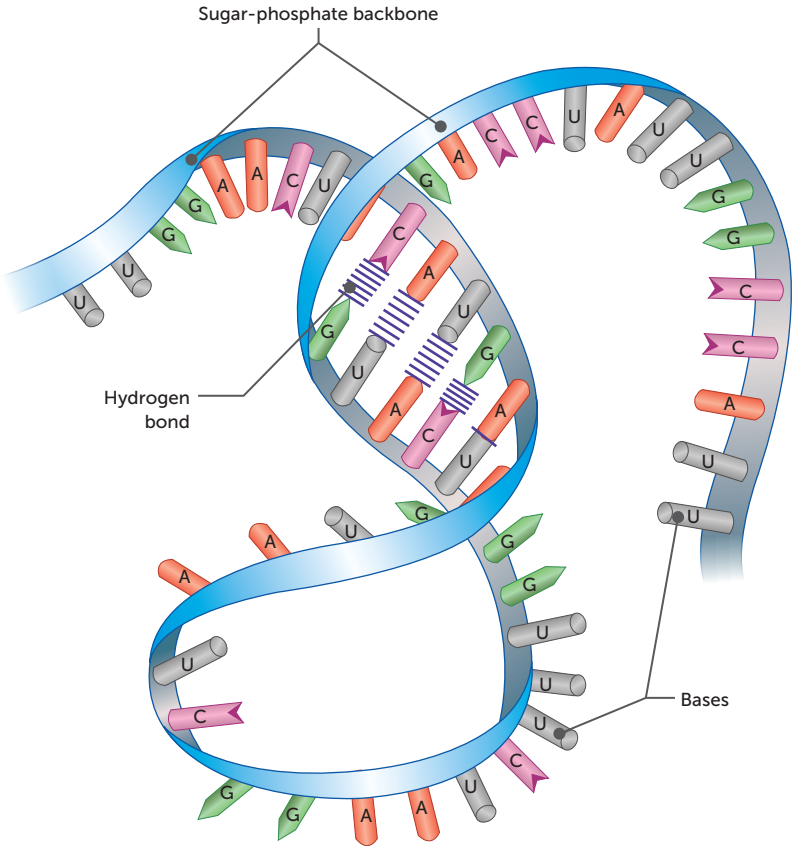


FIGURE 9.10 RNA is a single-stranded nucleic acid molecule

There are different types of RNA, each with its own role in protein synthesis.

- **Messenger RNA (mRNA)** is made in the nucleus and takes the genetic code into the cytoplasm allowing the genetic code to be 'read' by ribosomes.
- **Ribosomal RNA (rRNA)** makes up approximately 60% of the mass of ribosomes, with the other 40% being protein. The rRNA ensures the correct alignment of mRNA, tRNA and ribosome. It also has an enzymatic role in the formation of peptide bonds between amino acids.
- **Transfer RNA (tRNA)** is a small molecule of RNA, containing only 70–90 nucleotides. Each tRNA molecule is able to carry a specific amino acid and therefore plays a vital role in protein synthesis.

Key concept

Ribonucleic acid (RNA) is single-stranded nucleic acid that incorporates the bases adenine, cytosine, guanine and uracil. The types of RNA are messenger RNA, ribosomal RNA and transfer RNA.

From DNA to proteins

DNA contains the genetic code, the sequences of bases determining the proteins that are produced. A sequence of three bases is the code for a particular amino acid and is called a **triplet** (or a base triplet). For example, the sequence CAG (cytosine–adenine–guanine) is a DNA code for the amino acid valine; TTA is a code for leucine; and CCC is a code for proline. Thus, if the bases in part of a DNA molecule occurred in the order CAG TTA CCC, then the amino acids valine, leucine and proline would be assembled in that order in the protein made using instructions from that part of the DNA molecule.

9780170449090

In Chapter 2 you learnt that, while most DNA is found in the nucleus and is too large to leave, proteins are produced by ribosomes in the cytosol. This means that there must be processes to transfer the genetic code from the nucleus to the cytosol.

Transcription

Transcription is the process by which the genetic instructions are copied (or transcribed) from the DNA to the mRNA molecule. The transcription is triggered by chemical messengers that enter the nucleus from the cytosol and bind to the DNA at the relevant gene. This causes an enzyme called **RNA polymerase** to begin the process of making mRNA.

First, the helicase enzyme makes the double-stranded DNA molecule come apart. It usually separates about 17 base pairs at a time. RNA polymerase then transcribes (copies) the bases on one strand of the DNA to make a complementary molecule of mRNA.

- Where there is a cytosine base (C) on the DNA, a guanine (G) will be added to the mRNA.
- Where there is a guanine base (G) on the DNA, a cytosine (C) will be added to the mRNA.
- Where there is a thymine (T) on the DNA, an adenine (A) is added to the mRNA.
- Where there is an adenine (A) on the DNA, a uracil (U) is added to the mRNA.

Figure 9.11 shows the process of mRNA formation. At the end of the gene, a sequence of bases tells the RNA polymerase to stop copying, and as a result the mRNA molecule is released. Often other RNA polymerases will follow the first one, so that many copies of the same mRNA are made.

You can see in Figure 9.11 that only one of the strands of the DNA molecule is copied during transcription. The strand that is copied is called the **template strand** because it is the template from which the mRNA is made. The other DNA strand is known as the **coding strand**. Because the bases always form complementary pairs, the order of bases on the coding strand will be the same as in the mRNA molecule (except that uracil will be present in place of thymine).

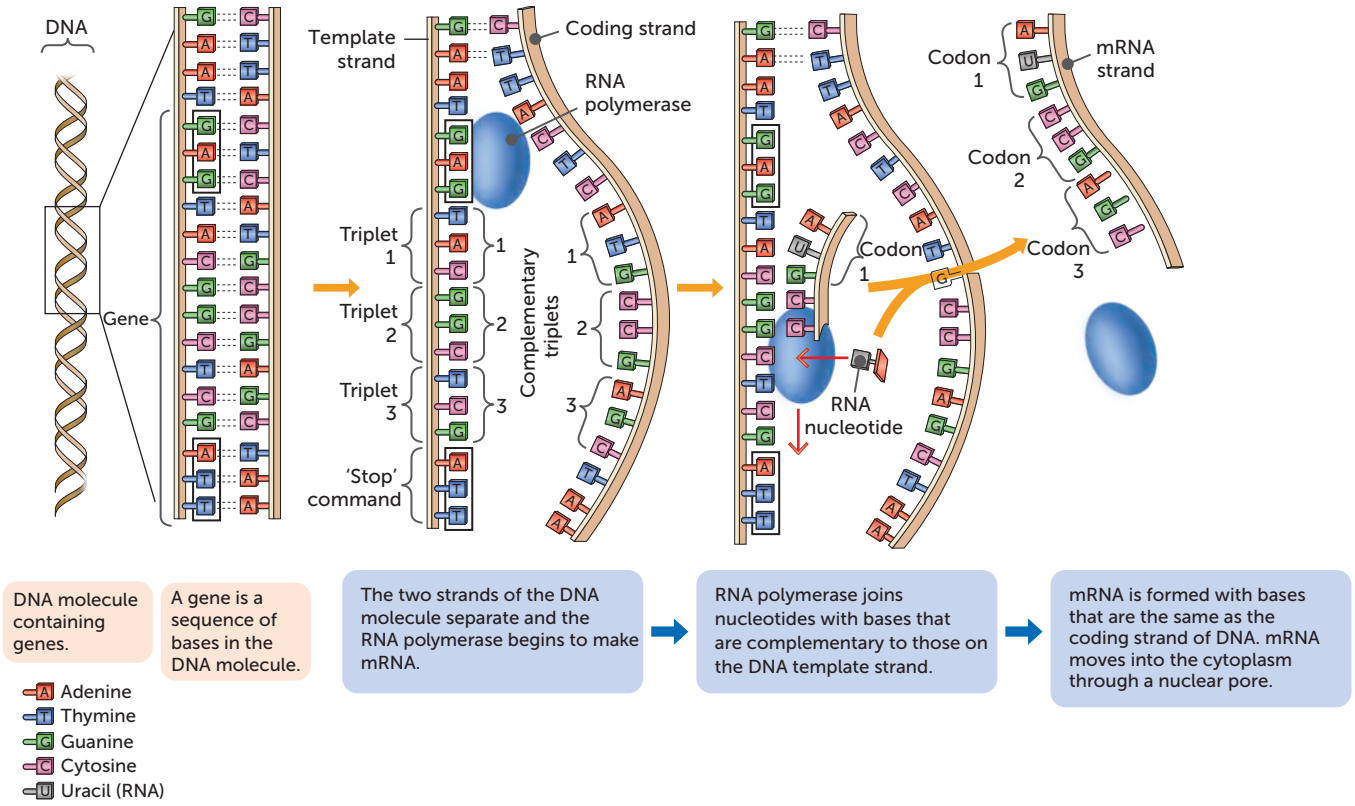


FIGURE 9.11 Transcription of the DNA code to form messenger RNA

Translation

Translation is the production of a protein using the information that is coded in the mRNA molecule. In the cytosol, a ribosome attaches to one end of the mRNA molecule at a particular sequence of bases (adenine, uracil, guanine) called the **start codon**. This ensures that the ribosome attaches to the correct end of the mRNA.

The ribosome then moves along the mRNA three bases at a time. Each group of three bases is called a **codon** and corresponds to a specific amino acid. The start codon, AUG, also codes for the amino acid methionine. This means that every protein begins with methionine when it is first made. However, it may be removed later.

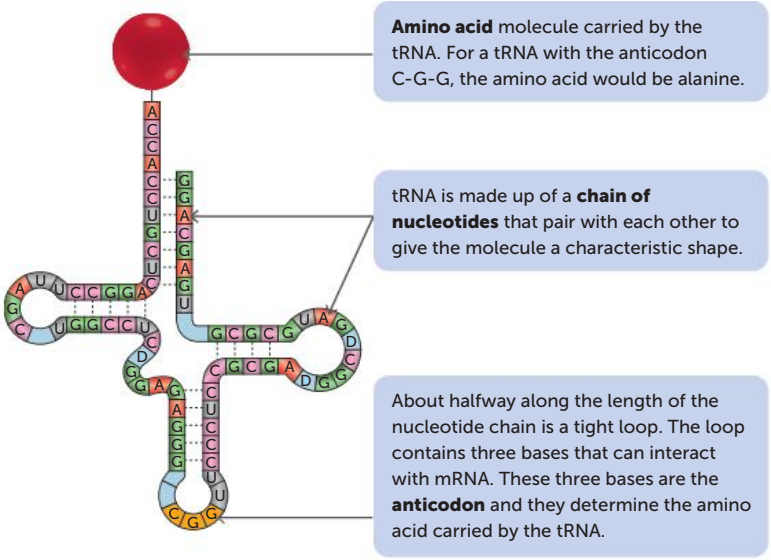


FIGURE 9.12 Structure of transfer RNA

As the ribosome reads the codons on the mRNA, the tRNA molecules with the complementary bases join to the mRNA. The sequence of three bases matching the codon is called the **anticodon**.

The amino acids carried by the tRNA are joined so that the protein is assembled with the amino acids in the correct sequence. For each bond formed between the amino acids, the energy from the breakdown of one ATP molecule is required. Once the tRNA has delivered its amino acid, it detaches from the ribosome and can then pick up another amino acid from the cytosol.

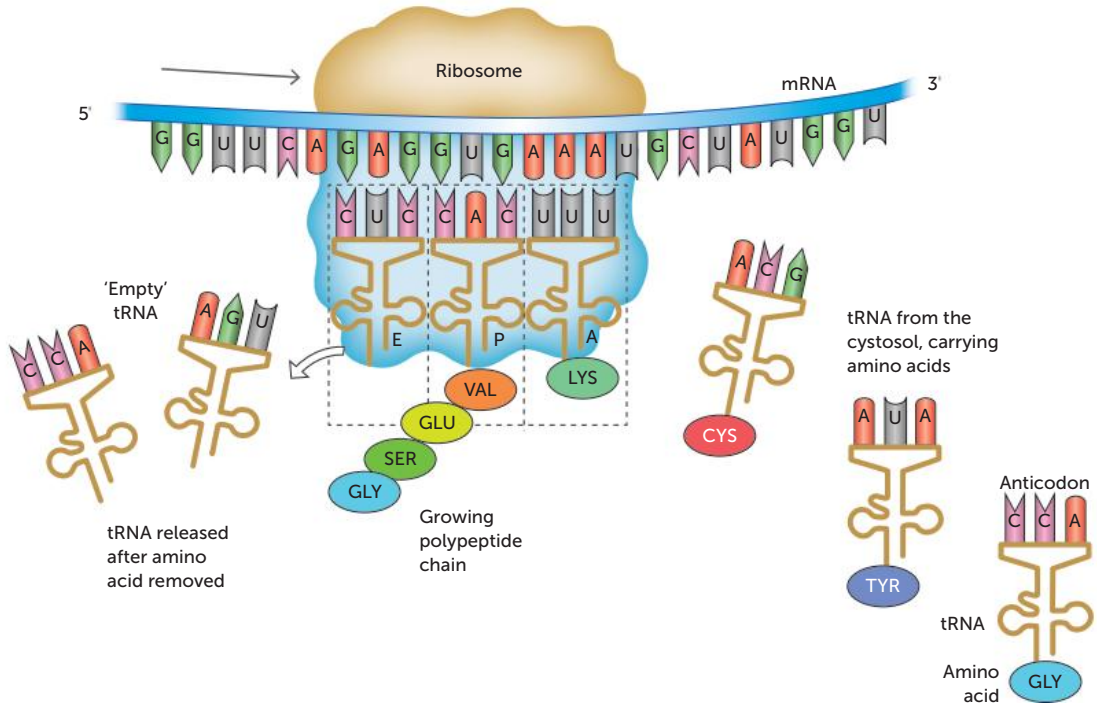


FIGURE 9.13 The anticodon on the tRNA molecule binds to the appropriate codon on the mRNA molecule, bringing the correct amino acid into the peptide chain

Key concept

The base sequence on the DNA is transcribed to a molecule of messenger RNA that is able to leave the nucleus. The mRNA binds to a ribosome, where it is translated by molecules of transfer RNA adding the appropriate amino acids to form the protein.

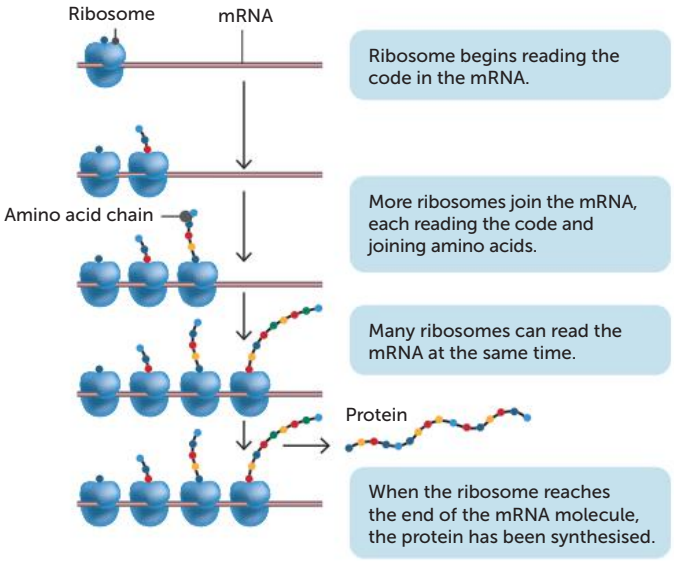


FIGURE 9.14 One mRNA molecule may be read by many ribosomes at the same time, producing multiple copies of the protein.

Gene expression

The process of copying information from DNA on to messenger RNA (mRNA) and then translating the message into a series of amino acids to form a protein is called **gene expression**. The genes contain the instructions for making mRNA, but at any given time a cell is making mRNA from only a fraction of its genes. For example, genes for the production of insulin are activated in some cells in the pancreas but not in bone or muscle cells. Genes that are being used to make mRNA are said to be 'switched on'; a gene not being used to make mRNA is 'switched off'. Many factors determine whether a gene is being expressed – that is, whether it is on or off. They include the age of the cell, time of day, signals from other cells, the environment of the cells, and whether or not the cell is dividing.

Although the process of gene expression seems very complicated, it all happens very quickly. One ribosome can make a protein consisting of 400 amino acids in about 20 seconds. One mRNA often has 10 or 20 ribosomes reading its code at the same time, and a cell may have 300 000 identical mRNA molecules. This means that a cell could produce over 150 000 protein molecules per second!

Gene expression
This website provides more on gene expression.

Protein synthesis
This website features an animation of protein synthesis.

Key concept

Only certain genes are transcribed in each cell. Genes are switched on or off in accordance with the type of cell and its activity.

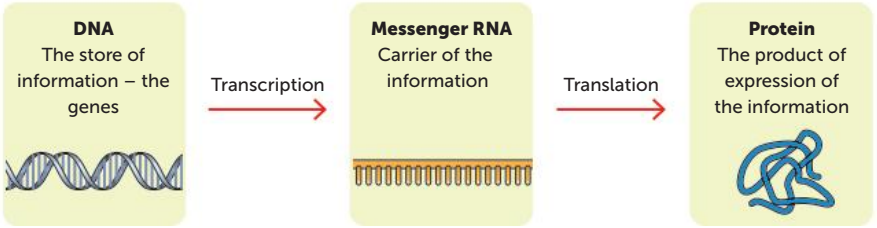


FIGURE 9.15 Simple summary of gene expression

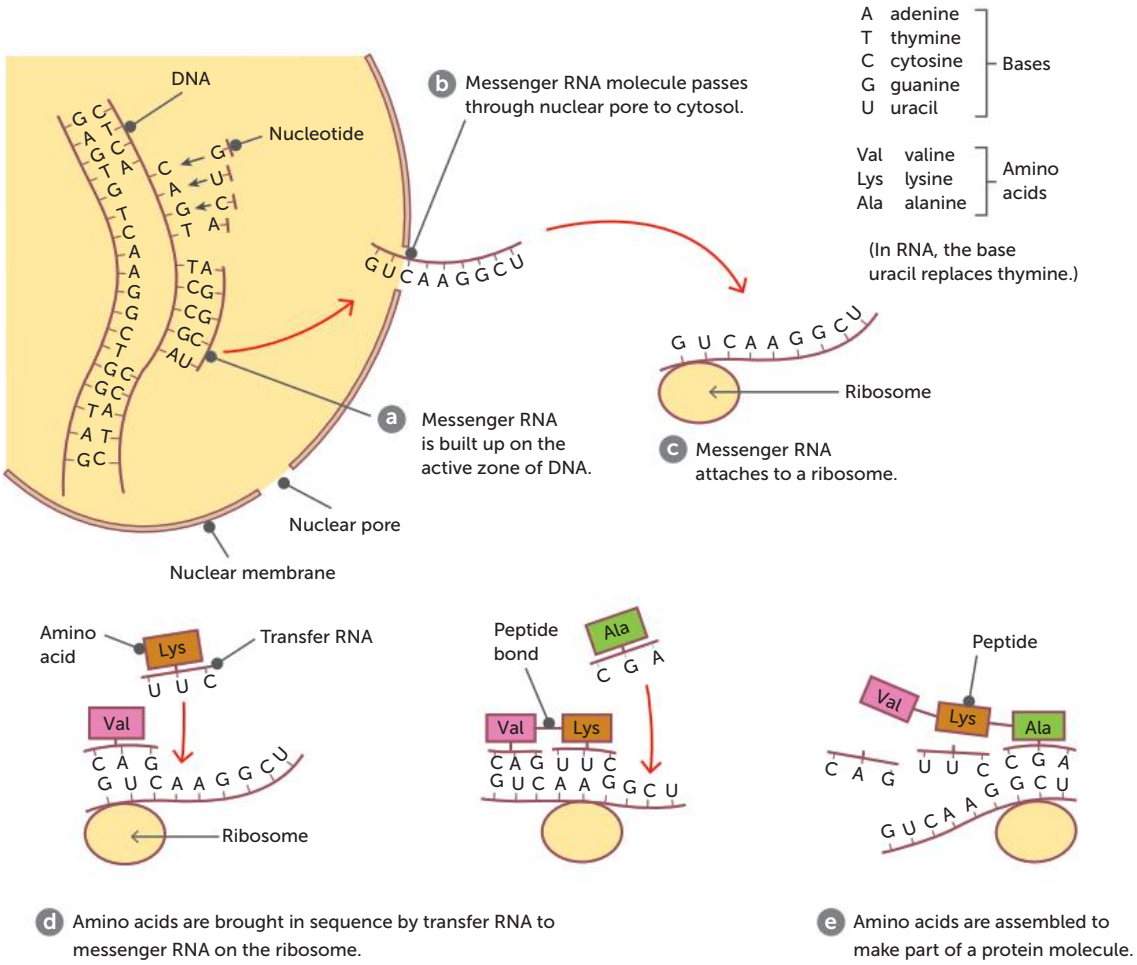


FIGURE 9.16
Synthesis of proteins from the DNA code

WS
9.2 Protein synthesis

Lipid and carbohydrate synthesis

There are no genes that carry instructions for the manufacture of lipids or carbohydrates. However, the synthesis of these substances requires enzymes, and enzymes are proteins. As we have seen, the DNA in the genes carries the code for protein manufacture. Thus, indirectly, the genes control the synthesis of lipids and carbohydrates.

TABLE 9.1 Types of nucleic acids

Deoxyribonucleic acid (DNA)	Very large molecule made of two strands of nucleotides that are joined by bonds between the nucleotide bases. The two strands are twisted into a double helix. Found in the nucleus and mitochondria of cells.
Nuclear DNA (nDNA)	DNA found in the nucleus of cells.
Mitochondrial DNA (mtDNA)	DNA found in the mitochondria.
Ribonucleic acid (RNA)	Large molecule composed of a single strand of nucleotides.
Messenger RNA (mRNA)	RNA molecule that carries the code for protein synthesis from the DNA in the nucleus to the ribosomes where the protein is made.
Transfer RNA (tRNA)	A small RNA molecule that transfers the correct amino acid to the ribosome for inclusion in the protein molecule being made.

9780170449090

Questions 9.2

RECALL KNOWLEDGE

1 List the types of RNA.

2 Complete the following table to compare DNA and RNA.

	DNA	RNA
Type of sugar		
Number of strands		
Bases		

3 The sequence of three bases in DNA, mRNA and tRNA determines the amino acid in a protein. State the name of the three bases in each of the molecules.

4 Name the enzyme that:
   a separates the strands of DNA
   b transcribes the bases on the DNA molecule to produce mRNA.

5 Describe the role of anticodons in the process of protein synthesis.

6 Describe the structure of mitochondrial DNA.

APPLY KNOWLEDGE

7 Explain why messenger RNA is needed for protein synthesis.

8 Explain why the base sequence of mRNA is the same as the coding strand.

9 Explain why gene expression must be controlled.

9.3 EPIGENETICS

Some of the factors that make genes more or less likely to be expressed may be inherited. Such factors are said to be **epigenetic**. *Epi* means 'on top of', or 'in addition to', so 'epigenetics' refers to changes in gene expression that result from mechanisms other than changes in the genes – that is, in the DNA or genome. Epigenetics is an important area of research where scientists try to find out the factors that help to make us who we are and influence what diseases we might acquire.

The study of epigenetics has changed the way we think about inheritance. It used to be thought that characteristics could only be passed on to the next generation through the genes. This meant that the environment a person was exposed to had no effect on their offspring. We now know that there is a growing list of environmental factors that can cause epigenetic changes that are inherited by offspring. In other words, what a person does during their lifetime may cause changes that are passed on to their children.

A person's genome is the hereditary information that is encoded in their DNA. Their epigenome is the sum of all the factors that determine when, where and which genes are 'switched on' or expressed. The epigenome helps to control which genes are active in a particular cell and therefore which proteins will be produced. Thus, epigenetic factors tell muscle cells to behave like muscle cells, nerve cells to act as nerve cells, and so on. If the epigenome is abnormal, certain cells may be abnormal and disease may result.

One way in which genes are regulated epigenetically is through changes in chromatin.

Chromatin

DNA molecules are in the form of long strands. Their length in a human cell is estimated to be between 2 and 3 metres. Cells are microscopic, so for 2–3 m of DNA to fit inside the nucleus of a cell, the DNA has to be tightly coiled. The DNA molecules are coiled around special proteins called histones. When a cell is not dividing, the coiled DNA forms a tangled network. The DNA and the histone proteins associated with it are called chromatin.

Gene expression may change if the way in which the DNA is wrapped around the histone changes. There are a number of ways in which such a change in the structure of the chromatin can occur.

If some of the amino acids that are in the histone proteins are changed, this will change the shape of the histone. The modified histone shape may be copied each time a new DNA molecule is formed.

9780170449090

The modified histone would ensure that a stem cell that differentiated into a liver cell would remain a liver cell and not revert to being a stem cell.

Another histone modification that may occur is **acetylation** – the addition of an acetyl group (CH_3CO) to the histone protein. This reduces the attraction between histones and DNA, relaxing the structure of the chromatin. This promotes transcription by allowing RNA polymerase access. Therefore, acetylation *enhances* gene expression.

Chromatin remodelling may also occur by adding methyl groups (CH_3) to the DNA molecule or histone proteins. DNA **methylation** usually occurs at sites on the DNA molecule where a cytosine nucleotide is adjacent to a guanine nucleotide. These are known as **CpG sites**, cytosine–phosphorous–guanine. Methylation of DNA *inhibits* gene expression by restricting access to RNA polymerase.

Histone methylation may either increase or decrease the transcription of genes, depending on where the methyl groups attach and how many become attached. If the methylation causes the chromatin structure to relax, it will increase transcription.

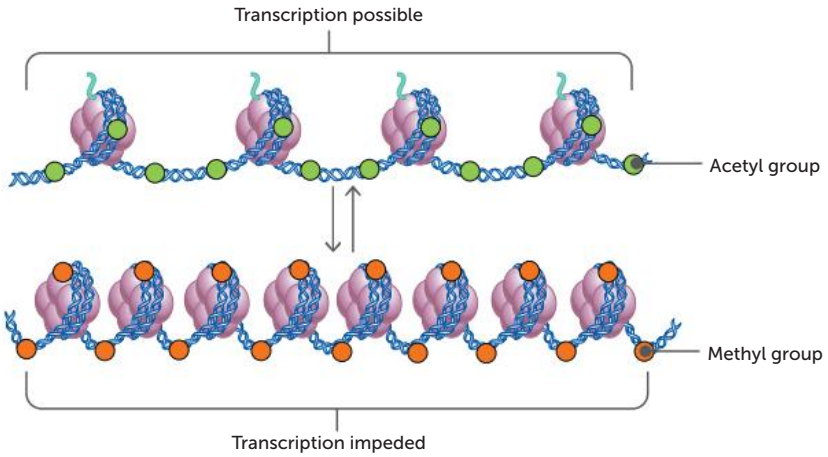


FIGURE 9.17
Acetylation relaxes the chromatin and increases transcription, whereas methylation tightens the chromatin, which decreases transcription

Environment and the epigenome

A person's epigenome can be changed by exposure to certain environmental stimuli. Examples of environmental agents that may cause epigenetic changes are severe stress, nutritional factors, and toxins or drugs that may enter cells. Such agents do not change the DNA, but they interfere with the transcription and translation processes involved in protein production. The mechanism by which the epigenome affects gene regulation is still under investigation, but it is known that epigenetic factors can influence any step in gene expression – any step in the pathway from gene to protein.

Key concept

Epigenetics is the study of variations that occur due to factors that switch genes on and off. This process includes modifying the shape of histone proteins, acetylation and methylation.

Questions 9.3

RECALL KNOWLEDGE

1 Define 'epigenetics'.
2 List the ways in which the structure of chromatin may be altered.
3 Describe acetylation.

APPLY KNOWLEDGE

4 Explain how the environment may affect gene expression.
5 Explain why gene expression increases when the chromatin structure is relaxed.
6 Explain why identical twins are used when studying epigenetics.
7 Compare and contrast acetylation and methylation.
8 Suggest why the number of epigenetic changes in our cells increases as we get older.

Epigenetics
This website provides information about the epigenome.

9780170449090

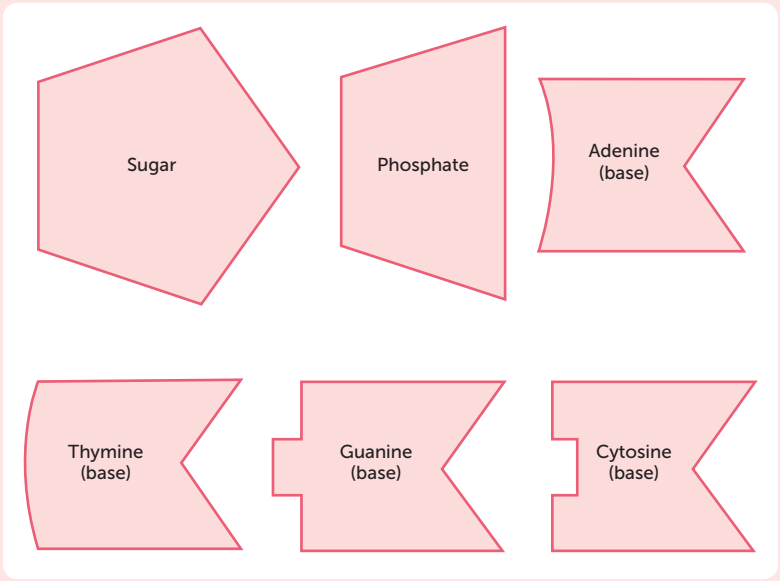
CHAPTER 9 ACTIVITIES

ACTIVITY 9.1 Modelling DNA structure and replication

A scientific model is a simplified representation of an idea or process. In this activity, you will build a simple model of a DNA molecule and use it to demonstrate how the model can form an exact replica of itself.

You will need

Shapes (cut out of cardboard or plastic) to represent sugar (x24), phosphate (x24), adenine (x6), thymine (x6), guanine (x6) and cytosine (x6); sticky tape. *Note*: The edges of the shapes must fit together perfectly for this activity, so keep this in mind when cutting them out. Suitable shapes are shown below.



What to do

DNA molecule

DNA is made up of thousands of units called nucleotides. Each nucleotide consists of a sugar (deoxyribose), a phosphate and a base containing the element nitrogen. The nucleotides are arranged so that the molecule has two frameworks of alternating sugars and phosphates. The two backbones are joined by the bases of the nucleotides.

1. Arrange the shapes to make at least 24 nucleotides. Use sticky tape to join these together.
2. Arrange 12 of the nucleotides to form the structure of a DNA molecule.
3. Using the letters A, T, G and C, write down the order of the base pairs in the molecule you have made (list the pairs from top to bottom).
4. Separate the DNA molecule that you have made into two halves by separating the two chains of nucleotides.
5. Use your remaining nucleotides to make new strands for each half. You may need to make more nucleotides if you do not have the bases required.
6. Write down the order of the base pairs in your two new molecules.

Genetic Science Learning Center: build DNA

Studying your observations

After completing this activity, answer the following questions:

1. How does the order of the base pairs of your two new molecules compare with the sequence of the original molecule? Explain why this happens.
2. Why are the numbers of adenine and thymine bases in a DNA molecule always equal?
3. Would there be equal numbers of thymine and cytosine bases? Explain your answer.
4. Explain how, when a cell divides, the two daughter cells contain the same genetic information as the parent cell.

ACTIVITY 9.2 Extracting DNA

In this activity, you will extract DNA from plant or animal material.

You will need

Plant or animal material such as wheatgerm, split peas, onion or chicken liver; food blender or processor; salt; liquid detergent; methylated spirits; fine kitchen sieve; clean beakers; wooden skewer

What to do

1. Place one-quarter of a cup (about 100 mL) of the plant or animal material in the blender.
2. Dissolve half a teaspoon of salt in 150 mL of warm water (about body temperature) and add the solution to the material in the blender.
3. Blend on high for about 30 seconds.
4. Separate the liquid from the solid material by pouring the contents of the blender into a fine kitchen sieve and collecting the liquid in a 250 mL beaker.
5. Add a teaspoon of liquid detergent and stir gently every minute for 5 minutes.
6. Tilt the beaker and slowly pour methylated spirits down the side until it forms a 1 cm layer on top of the solution in the beaker. Make sure the methylated spirits does not mix with the solution.
7. Allow the mixture to stand and you should see white, stringy DNA form at the interface between the watery solution and the methylated spirits.
8. Twist a wooden skewer in the layer of DNA and you may be able to lift some of it out of the solution.

For further investigation

Design investigations to determine:

- if all plant or animal material has DNA that appears the same
- if you can extract more DNA from some plant or animal materials than from others.

You may be able to carry out your investigations by using fruit such as bananas, vegetables such as silver beet, meat, seaweed or mushrooms.

9780170449090

CHAPTER 9 SUMMARY

- DNA, or deoxyribonucleic acid, is a polymer made up of many nucleotides joined together in two strands that twist to made a double helix.
- Each nucleotide has a sugar molecule, a phosphate group and one of the nitrogenous bases – adenine, thymine, guanine or cytosine.
- The genetic code is the order of bases in DNA.
- DNA is wrapped around eight proteins called histones, forming a nucleosome.
- When a cell divides, the length of DNA coils to form a chromosome.
- During cell division, DNA must replicate by the strands separating and acting as a template for nucleotides to join and form a complementary strand.
- Sections of DNA on chromosomes contain the genetic code for the production of proteins.
- RNA, or ribonucleic acid, is a single-stranded nucleic acid that differs from DNA, as its sugar is ribose and it contains uracil instead of thymine.
- RNA may be messenger RNA, transfer RNA or ribosomal RNA.
- A series of two bases coding for an amino acid is called a triplet on DNA, a codon on mRNA, and an anticodon on tRNA.
- In transcription, a molecule of mRNA is produced that is complementary to the template strand of DNA.
- mRNA is small enough to leave the nucleus through the nuclear pores.
- In translation, the mRNA attaches to a ribosome and facilitates complementary tRNA molecules that bring the corresponding amino acids to form a protein molecule.
- Gene expression is the process of converting instructions in our DNA into functional products.
- Gene expression is regulated to control what each cell is producing at any time.
- DNA regulates lipid and carbohydrate production through the production of enzymes.
- Mitochondrial DNA is a circular molecule with no associated proteins. It is important for normal mitochondrial functioning.
- Epigenetics relates to the chemical reactions that activate or deactivate the genome – in particular, the changes in the chromatin (the DNA and associated histones).
- Chromatin changes come about from modification of the shape of histone proteins, the addition of acetyl groups to the histones, and the methylation of either DNA or histones.

9780170449090

CHAPTER 9 GLOSSARY

Acetylation Addition of an acetyl group to a histone protein so that gene expression is enhanced

Adenine One of the nitrogenous bases found in DNA and RNA

Anticodon A sequence of three bases in a tRNA molecule that binds to a complementary sequence of three bases (the codon) in an mRNA molecule, to specify a particular amino acid during protein synthesis

Chromatin A tangled network of DNA in the nucleus of a cell that is not dividing

Chromosome One of the 46 rod-like structures that appear in the nucleus of a human cell at the commencement of cell division

Coding strand The strand of a DNA molecule that is not used to form mRNA

Codon The strand of a DNA molecule that is not used to form mRNA

CpG site A site on a DNA molecule where a cytosine nucleotide is next to a guanine nucleotide; DNA methylation usually occurs at these sites

Cytosine One of the nitrogenous bases found in DNA and RNA

Deoxyribonucleic acid (DNA) A molecule in the nucleus of a cell that determines the types of protein a cell can make

DNA ligase Enzyme that joins short sections of DNA together

DNA polymerase Enzyme that joins nucleotides together

DNA replication The production of an identical copy of the DNA

Double helix The spiral shape of the DNA molecule

Epigenetics Altering expression of a gene without changing the gene structure

Gene Section of a chromosome that contains the nucleotide sequence coding for a particular trait

Gene expression The process where information in a gene is used to make a product

Guanine One of the nitrogenous bases found in DNA and RNA

Helicase The enzyme responsible for the separation of DNA strands during replication

Histone A special protein around which DNA is coiled to form chromatin

Messenger RNA (mRNA) An RNA molecule that transfers coded information from the nucleus to the ribosomes

Methylation The addition of a methyl group to a DNA molecule that results in inhibition of gene expression

Mitochondrial DNA (mtDNA) DNA found in the mitochondria

Nuclear DNA DNA found in the nucleus

Nucleosome A length of DNA wound around eight histone proteins

Nucleotide Units of phosphate, sugar and nitrogen base that make up the DNA molecule

Ribonucleic acid (RNA) A special molecule that takes the code for amino acids from DNA to the ribosomes

Ribosomal RNA (rRNA) Nucleic acids that form part of the ribosomes

RNA polymerase An enzyme that is active during transcription of RNA from DNA

Start codon The sequence AUG (adenine, uracil, guanine) is the codon for the amino acid methionine; when a ribosome reaches this codon, it starts making protein

Template strand The strand of a DNA molecule that is used to form the sequence of bases in mRNA and is copied during transcription

Thymine One of the nitrogenous bases found in DNA

Transcription The process by which messenger RNA is formed, using the code in a DNA molecule by matching the sequence of nucleotides

Transfer RNA (tRNA) An RNA molecule that brings amino acids from the cytoplasm to the ribosomes

Translation The production of a protein using the information coded in the mRNA molecule; another name for protein synthesis

Triplet A sequence of three bases that is the code for a particular amino acid; also called a base triplet

Uracil One of the nitrogenous bases found in RNA

9780170449090

CHAPTER 9 REVIEW QUESTIONS

Recall

1 Describe the difference between a gene and a chromosome.
2 Draw a labelled diagram to illustrate the structure of DNA.
3 List the nitrogenous bases found in RNA.
4 Describe the role of the ribosomes in protein synthesis.
5 Define 'gene expression'.
6 List the functions of mitochondrial DNA.

Explain

7 Explain the difference between:
  a DNA and RNA
  b messenger RNA and transfer RNA
  c transcription and translation.
8 Explain how a DNA molecule is able to form an exact replica of itself.
9 Explain the role of mRNA in protein synthesis.
10 How does DNA control the synthesis of carbohydrates and lipids in a cell?
11 Histones affect both the structure and function of DNA. Explain how they achieve each of these effects.

Apply

12 Explain how a person's genome is different from their epigenome.
13 The table below shows the results of analysing the nucleotides found in the cells of humans, chickens and wheat plants. What do these percentages tell us about the structure of DNA?

CELLS	PERCENTAGE OF NUCLEOTIDES CONTAINING:			
	ADENINE	GUANINE	CYTOSINE	THYMINE
Human	30.9	19.9	19.8	29.4
Chicken	28.8	20.5	21.5	29.2
Wheat	27.3	22.7	22.8	27.2

14 Approximately how many bases would there be in a messenger RNA molecule that coded for a protein 250 amino acids long? Explain your answer.
15 A DNA molecule has the following sequence of bases on the template strand: CTC CCC TTA GTC GAT AGT.
  a What would be the sequence of bases on the coding strand of the DNA molecule?
  b What sequence of bases would be found in a strand of messenger RNA?
16 Explain how the discovery of epigenetic factors has changed the way we think about characteristics that can be passed from one generation to another.

9780170449090

Extend

17 Geneticist Danielle Reed from the Monell Chemical Senses Center in Philadelphia, Pennsylvania, has said: 'Things written in pen you can't change. That's DNA. Things written in pencil you can. That's epigenetics.' Explain what you think Reed meant by this statement.

18 Gene expression is a complex process, far more involved than the simple explanation given in this chapter. Use the Internet to research the control of gene expression. In your research, you may come across the following terms: enhancers, insulators, operators, promoters, repressors and silencers. Find out how these various elements are involved in the process of gene expression. Use a table to summarise your findings.

19 Antibiotics, such as tetracycline, streptomycin and erythromycin, work by blocking translation during the process of protein synthesis. Suggest how this stops the bacteria replicating.

9780170449090

10

CELLS DIVIDE FOR GROWTH, REPAIR, REPLACEMENT AND REPRODUCTION

UNIT 2 CONTENT

SCIENCE INQUIRY SKILLS

- conduct investigations safely, competently and methodically for the collection of valid and reliable data
- interpret a range of scientific and media texts, and evaluate processes, claims and conclusions by considering the quality of available evidence; and use reasoning to construct scientific arguments

SCIENCE AS A HUMAN ENDEAVOUR

- new technologies, including Pap smear, breast screening and blood tests for prostate cancer, have made early detection of cancers possible

SCIENCE UNDERSTANDING

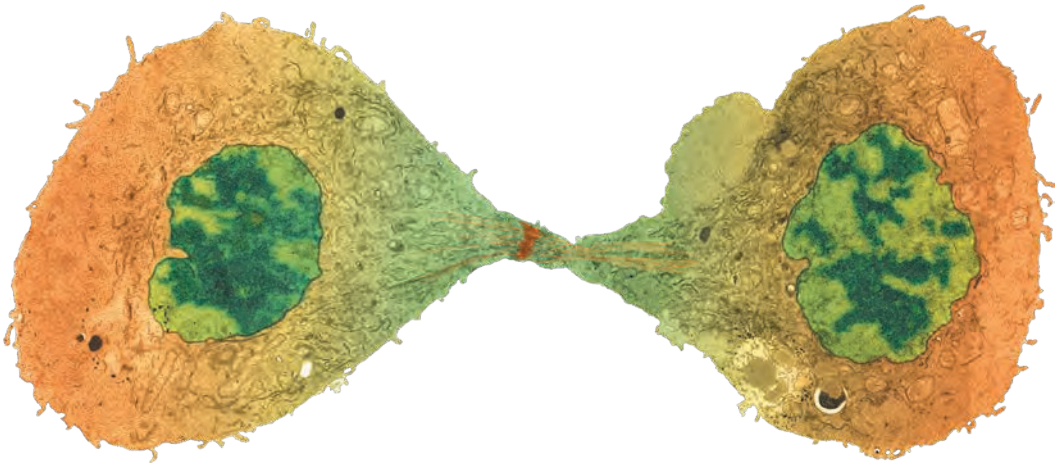
Cell reproduction

- mitosis forms part of the cell cycle producing new cells with the same genetic content
- the sequence of DNA replication, chromosome duplication and chromosome separation are important processes in the production of identical daughter cells by mitosis for growth, repair and replacement of tissues within the body
- stem cells have the ability to divide by mitosis and differentiate into many different tissues, depending on the level of cell potency
- uncontrolled division of cells can result in the development of tumours/cancers
- meiosis produces gametes for reproduction and involves DNA replication, chromosome pairing, and two successive nuclear divisions distributing haploid sets of chromosomes to each gamete
- crossing over, non-disjunction and random assortment of chromosomes during meiosis will produce gametes with different genetic content
- differences between mitosis and meiosis reflect their roles in the body

Source: School Curriculum and Standards Authority, Government of Western Australia

9780170449090

You have increased in size a lot since you were born. This growth was not due to an increase in the size of your cells, but rather an increase in the number of cells. New cells are constantly needed for growth and to replace cells that have died or been damaged. In this chapter, we examine the processes of producing new cells.



Science Photo Library/GSchmeissner

FIGURE 10.1 New cells are formed when one cell divides into two

10.1 THE CELL CYCLE

Cells need to reproduce for a number of reasons. When organisms grow, it isn't because the cells get larger; it is because they are made up of more cells. Therefore, more cells need to be produced. New cells are also needed to replace old, dead or damaged cells. Some human cells have a very short life span. For example, the cells lining the intestines live for less than two days. On the other hand, many nerve cells in the brain last a lifetime. Generally, the more wear and tear on a cell, the shorter the life span. Table 10.1 shows the life span of different types of human cells.

TABLE 10.1 Average life span of human cells

CELL TYPE	AVERAGE LIFE SPAN (DAYS)
Intestinal lining	1.3
Stomach lining	2.9
Tongue surface	3.5
Cervix (neck of the uterus)	5.7
Cornea of the eye	7.0
Outer skin of the abdomen	7.0
Inside of the cheek	10.0
Alveolus (air sac in the lung)	21.0
White blood cell	Depending on type and activity, from minutes to years
Red blood cell	120.0
Kidney	170.0
Bladder lining	330.0
Liver	450.0
Nerve cell in brain	29 200+ (80+ years)

9780170449090

The cell cycle

The events that take place from one cell division to the next are called the **cell cycle**. It is called a cycle because the events keep repeating as the cell divides again and again.

The events that occur in the cell cycle have been divided into a number of phases:

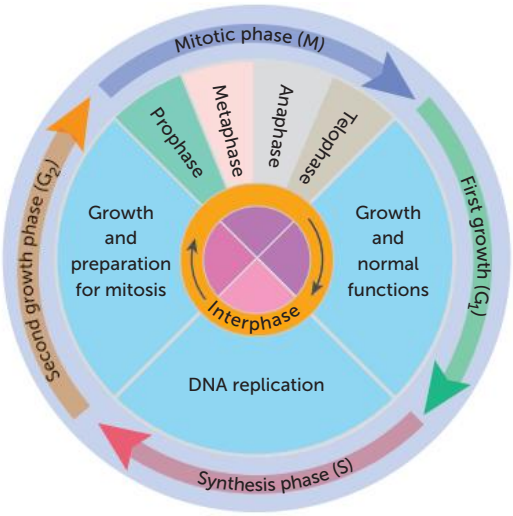


FIGURE 10.2 The cell cycle

- **G_1 phase**, or first growth phase – the cell produces new proteins, grows and carries out its normal tasks for the body; this phase ends when the cell's DNA begins to duplicate.
- **S phase**, or synthesis phase – the DNA molecules in the cell nucleus form exact copies of themselves.
- **G_2 phase**, or second growth phase – this relatively short phase involves preparation for cell division.
- **M phase**, or mitotic phase – the cell divides into two daughter cells.

After division, cells may continue the cycle and re-enter the G_1 phase. Some cells leave the cycle and stop dividing for days, years or even for the rest of the person's life. These cells are in the **G_0 phase**.

Mitosis

As you learnt in Chapter 9, DNA controls the structure and actions of cells, and hence the body. Therefore, it is vital that, when a cell reproduces, each new cell gets exactly the same DNA as the parent cell. In other words, each new cell must contain the same genetic information as the parent cell. This is achieved by division of the nucleus, known as **mitosis**. Mitosis ensures that each body cell receives the exact same hereditary material (DNA) as that possessed by its parent cell.

For convenience, biologists describe mitosis in four stages: prophase, metaphase, anaphase and telophase. However, the process is continuous; it does not occur in steps.

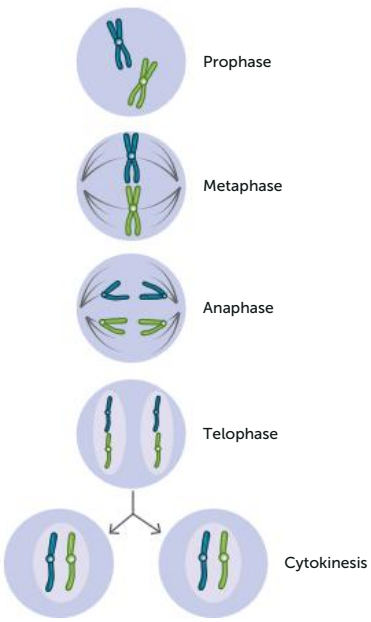


FIGURE 10.3 The stages of mitosis in a cell with two chromosomes

Interphase

Interphase is the period between nuclear divisions. During interphase, the cell goes through the G_1, S and G_2 phases of the cell cycle. In the S phase, the DNA molecules in the nucleus form exact copies of themselves. Thus, in the period between one cell division and the next, the quantity of DNA in the nucleus doubles.

Prophase

Prophase is the first phase of mitosis. Two pairs of centrioles become visible early in prophase. They move to opposite ends (or poles) of the cell and microtubules begin to radiate from them. At the same time, the nucleolus disappears and the nuclear membrane begins to break down. The chromatin threads of DNA become tightly coiled and can be seen as chromosomes. Coiling the long, delicate DNA molecules makes it easier to distribute the DNA to the daughter cells.

9780170449090

Each chromosome consists of two chromatids, which are joined at a point called the centromere. The two chromatids are identical, tightly coiled DNA molecules produced from DNA replication during interphase.

By the end of prophase, the centrioles have reached opposite poles of the cell and some of the microtubules radiating from them join to form a framework of fibres called a spindle. The nuclear membrane has now completely disappeared, and the chromatid pairs migrate towards the centre (equator) of the cell.

Metaphase

During **metaphase**, the chromatid pairs line up on the equator of the spindle. The centromere of each pair is attached to a spindle fibre.

Anaphase

In **anaphase**, each pair of chromatids separates at the centromere. As the chromatids have become independent of each other, they are now each called chromosomes. The new chromosomes are then pulled away from one another towards opposite poles of the cell. The centromeres are still attached to the spindle fibres, and it seems that the spindle fibres pull the chromosomes apart in some way.

Telophase

During **telophase**, the two sets of chromosomes form tight groups at each pole of the cell. A nuclear membrane forms around each group, and a nucleolus appears in each new nucleus. The spindle fibres disappear, and the chromosomes gradually uncoil to become chromatin threads once more.

Cytokinesis

Telophase is the last phase of nuclear division, but while the events of telophase are occurring, the cytoplasm usually begins to divide. Division of the cytoplasm is called **cytokinesis**. A furrow develops in the cytoplasm between the two nuclei. The furrow gradually deepens until it cuts the cytoplasm into two parts, each with its own nucleus. (*Note:* Although the term 'mitosis' is commonly used to refer to cell division, it technically refers just to the division of the nucleus.)

Mitosis and cytoplasmic division result in the formation of two daughter cells, which are now in interphase. Because each chromosome was duplicated prior to mitosis, and a copy went into each daughter cell, each daughter cell has exactly the same number and type of chromosomes as the parent cell. The genetic information is therefore passed from parent cell to daughter cells and without change.

Key concept

Mitosis results in the production of two daughter cells, each with an exact copy of the genetic information in the parent cell.

TABLE 10.2 Summary of cell division

STAGE	EVENTS OCCURRING
Interphase	DNA molecules duplicate.
Prophase	Nucleoli disappear; nuclear membrane breaks down; centrioles migrate to opposite poles; chromosomes appear as pairs of chromatids; spindle forms.
Metaphase	Chromosomes line up at the equator of the cell attached to the spindle.
Anaphase	Centromeres divide; chromosomes move to opposite ends of the spindle.
Telophase	Spindle disappears; nuclear membranes and nucleoli form; centrioles divide; chromosomes uncoil and disappear; cytokinesis begins.
Cytokinesis	Cytoplasm of the cell divides into two, each with a nucleus.

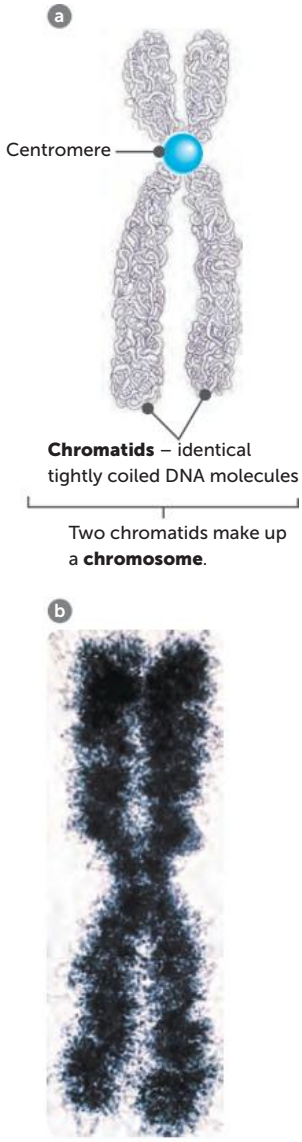
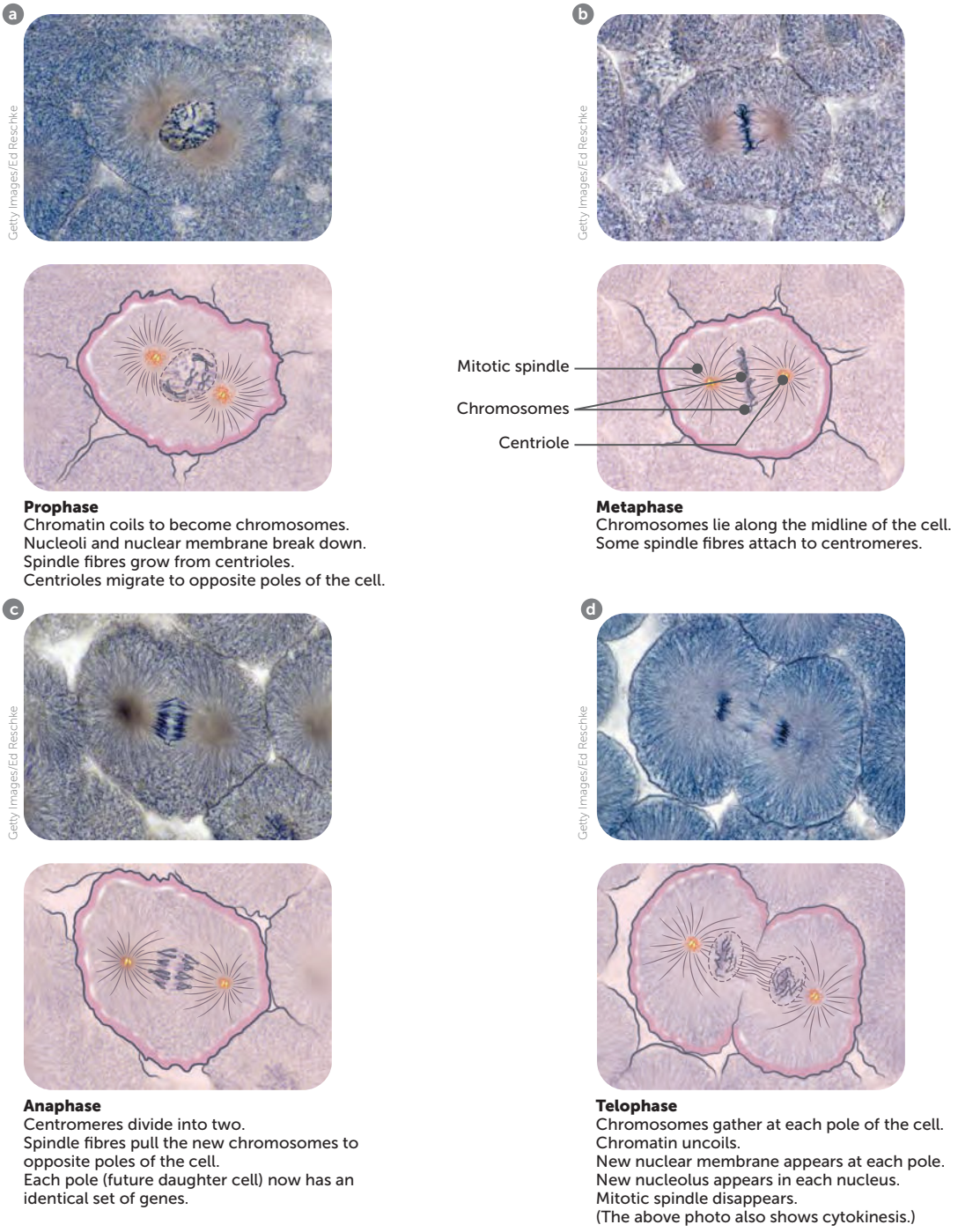


FIGURE 10.4 During prophase, chromatin threads become visible as chromosomes composed of pairs of chromatids. **a** Diagram of a chromosome; **b** Scanning electron micrograph of a chromosome

WS
10.1 Mitosis

9780170449090



Prophase
Chromatin coils to become chromosomes.
Nucleoli and nuclear membrane break down.
Spindle fibres grow from centrioles.
Centrioles migrate to opposite poles of the cell.

Metaphase
Chromosomes lie along the midline of the cell.
Some spindle fibres attach to centromeres.

Anaphase
Centromeres divide into two.
Spindle fibres pull the new chromosomes to opposite poles of the cell.
Each pole (future daughter cell) now has an identical set of genes.

Telophase
Chromosomes gather at each pole of the cell.
Chromatin uncoils.
New nuclear membrane appears at each pole.
New nucleolus appears in each nucleus.
Mitotic spindle disappears.
(The above photo also shows cytokinesis.)

Activity 10.1
Modelling mitosis and cytokinesis

Activity 10.2
Observing mitosis

FIGURE 10.5 The phases of mitosis: **a** prophase; **b** metaphase; **c** anaphase; **d** telophase. The drawing below each photograph shows a cell with only four chromosomes. Human cells have 46 chromosomes

Stem cells and differentiation

Cell differentiation
See a video of cell differentiation.

Cells can be classified as either stem cells or specialised cells. In general, specialised cells are unable to divide. Therefore, they must be replaced by other means. When stem cells undergo mitosis, the daughter cells may be new stem cells (stem cell proliferation) or cells that differentiate to form specialised cells.

Differentiation

Mitosis ensures that each daughter cell receives the same genes that were in the parent cell. Therefore, every cell in a person's body has the same genetic information. However, as seen in Chapter 2, cells are specialised so that they can carry out particular tasks. The process by which cells become specialised is called **differentiation**. It seems that as the cells undergo division by mitosis, different genes become activated. This makes the cells differentiate into specialised cells that can perform particular functions – for example, stomach cells that secrete enzymes, muscle cells that can contract, or red blood cells that can carry oxygen.

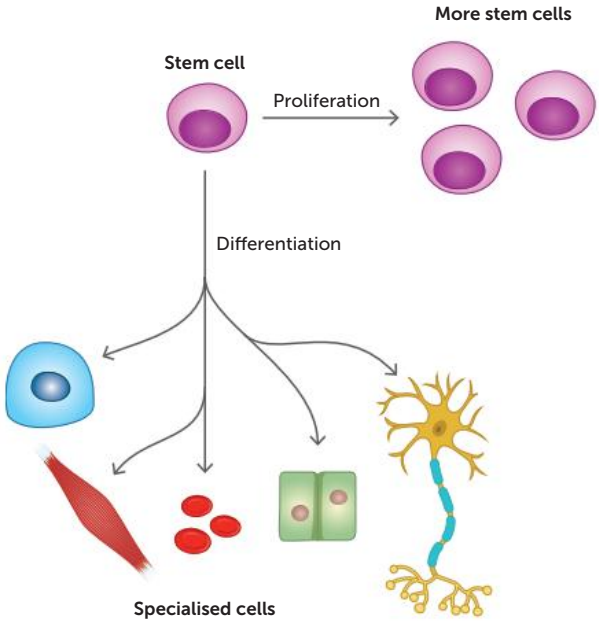


FIGURE 10.6 Stem cells can proliferate or differentiate to form specialised cells

Stem cells

The cells that can undergo differentiation are called **stem cells**. They are very different from other cells because they are not specialised for any particular role and are capable of repeated division by mitosis. In the right conditions, stem cells can differentiate into specialised cells. Because stem cells have the potential to develop into any cell type, they could possibly provide an unlimited source of cells for repair of tissues such as bone, skin, muscle, liver or blood.

Stem cells can be classified based on where they originate (embryonic, adult or cord blood) or the type of cells that they can form.

- **Totipotent stem cells** have the potential to create any type of cell necessary for embryonic development, including the embryo itself and all the membranes associated with embryonic development. The cells of the embryo within the first couple of mitotic divisions after fertilisation are the only totipotent cells.

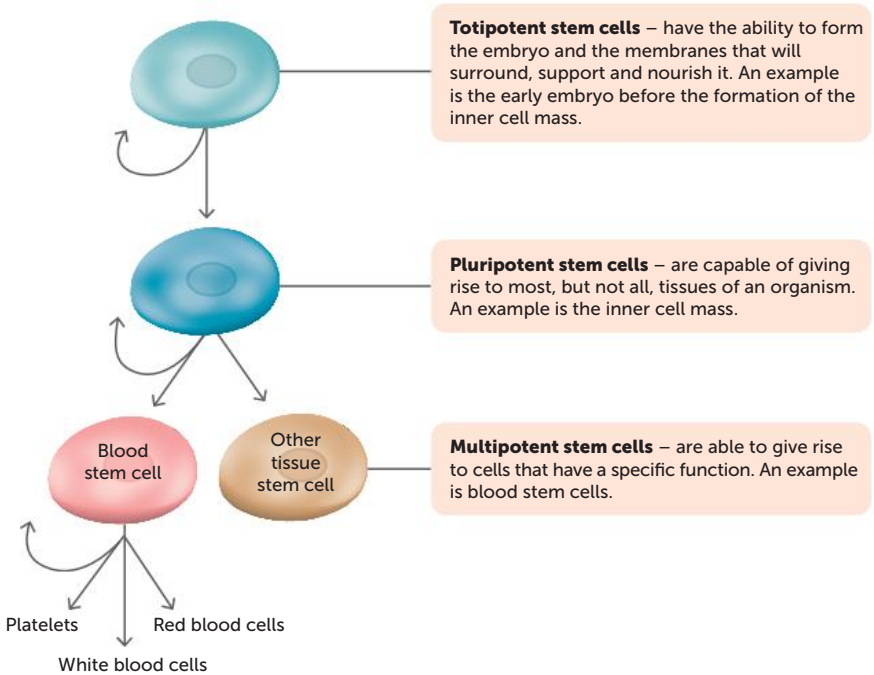


FIGURE 10.7 Types of stem cells and differentiation

- **Pluripotent stem cells** can give rise to any of the cells in the body. Embryonic stem cells are pluripotent as they differentiate to form all cells of the individual.
- **Multipotent stem cells** have the potential to form a number of different types of cells. For example, blood stem cells give rise to red blood cells, white blood cells and platelets, whereas skin stem cells give rise to the different types of skin cells. Embryonic stem cells, adult stem cells and cord blood stem cells are multipotent.

Stem cells
These websites feature extra information on stem cells.

Meiosis animation

Key concept

Stem cells have the ability to form new stem cells or to differentiate, forming specialised cells.

Questions 10.1

RECALL KNOWLEDGE

1. List the reasons that cells need to divide.
2. Place the stages of the cell cycle in order: metaphase, G1, G2, telophase, anaphase, S, prophase.
3. Name the stage where each of the following occurs:
   a. The chromosomes line up on the equator.
   b. The DNA replicates.
   c. Chromatin condenses and becomes visible.
   d. The nuclear membrane forms.
   e. Chromosomes move to opposite sides of the cell.
   f. The nuclear membrane disappears.
4. List the types of stem cells.

APPLY KNOWLEDGE

5. Explain why liver cells live for approximately 450 days, while cells lining the stomach live for only 2.9 days.
6. Explain why interphase was not listed in Question 2.
7. Contrast mitosis and cytokinesis.
8. Explain why it is important that embryonic cells from the first few cell divisions are totipotent.

10.2 PRODUCING GAMETES

The **gametes**, sperm and ova that are produced in the ovaries and testes, are the result of a special type of cell division called **meiosis**. Cells that make up the human body contain 46 chromosomes, and all cells that arise by the process of mitosis contain 46 chromosomes as well. If human gametes were produced by mitosis they also would contain 46 chromosomes. At **fertilisation**, the fusion of the sperm and egg results in a doubling of the chromosome number. Thus, the new individual would have 92 chromosomes. Such a person would produce sperm and eggs containing 92 chromosomes, and any resultant offspring would have 184 chromosomes. Therefore, if mitosis were responsible for the production of the gametes, the chromosome number would double with each succeeding generation. Obviously, this is not what happens. Instead, meiosis results in daughter cells with half the number of chromosomes that were present in the original cell. This is known as the **haploid number**, whereas the number of chromosomes in body cells is the **diploid number**. The chromosomes in diploid cells actually exist in pairs that are identical in shape and carry genetic information that influences the same characteristics. These are called **homologous chromosomes**.

When a cell has two of each type of chromosome, it has the diploid chromosome number. Diploid cells are designated *2n*, where *n* stands for the number of different types of chromosomes. The diploid number for humans is 46. In gametes, only *one* of each type of chromosome is present, or *n*, and therefore they are described as haploid. The haploid number for humans is therefore 23.

The process of meiosis involves two nuclear divisions, but the chromosomes only duplicate once.

- *Interphase:* Prior to undergoing meiosis, a cell goes through an interphase stage where it grows and the DNA is replicated. This DNA replication occurs in the same way as it does in mitosis.

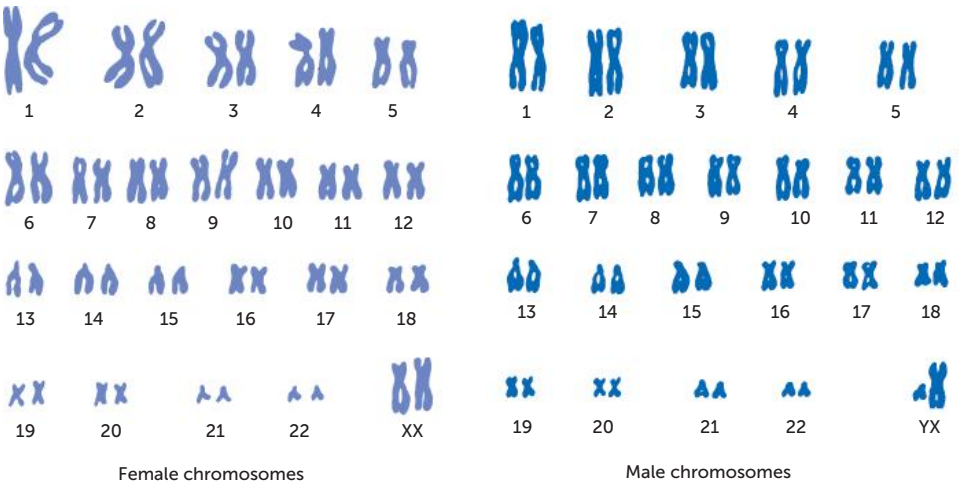


FIGURE 10.8 Human cells, except for gametes, are diploid and contain two chromosomes from each homologous pair

- *First division:* The homologous pairs separate and two daughter cells form with 23 chromosomes, each with two chromatids.
- *Second division:* The chromatids separate, resulting in four daughter cells with 23 chromosomes, each with one chromatid.

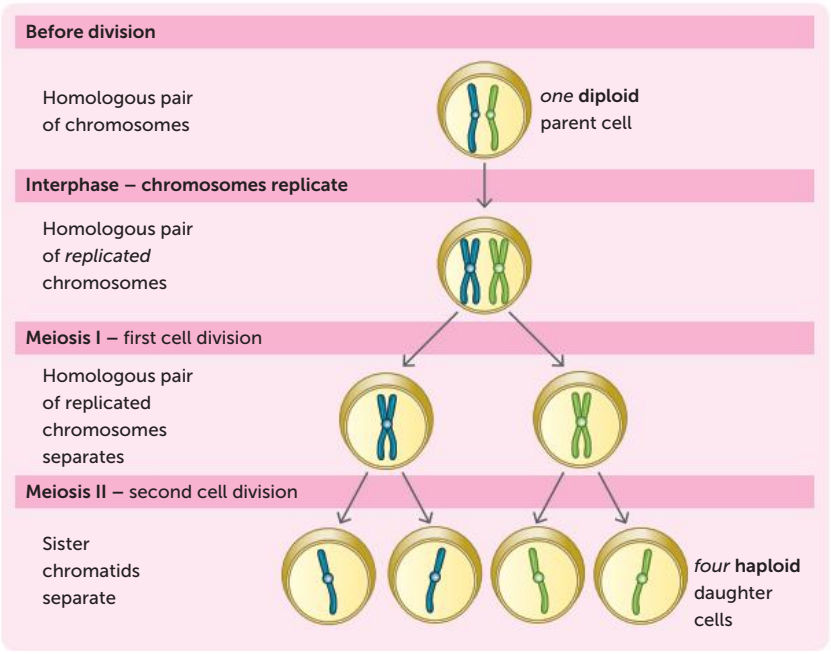


FIGURE 10.9 Summary of meiosis: one diploid parent cell produces four haploid daughter cells

First meiotic division

The first stage of meiosis starts with the chromosomes pairing off during the prophase stage. During **prophase I** of the first division of meiosis, the chromosomes become visible as long threads. Each has already undergone DNA replication and, therefore, consists of a pair of chromatids. These chromosomes gradually move, so that the members of a pair of homologous chromosomes come to lie alongside each other throughout their entire length. The chromosomes then shrink and thicken as the DNA becomes more tightly coiled. As each chromosome consists of two chromatids, each chromosome pair appears as *four* strands which frequently twist together.

9780170449090

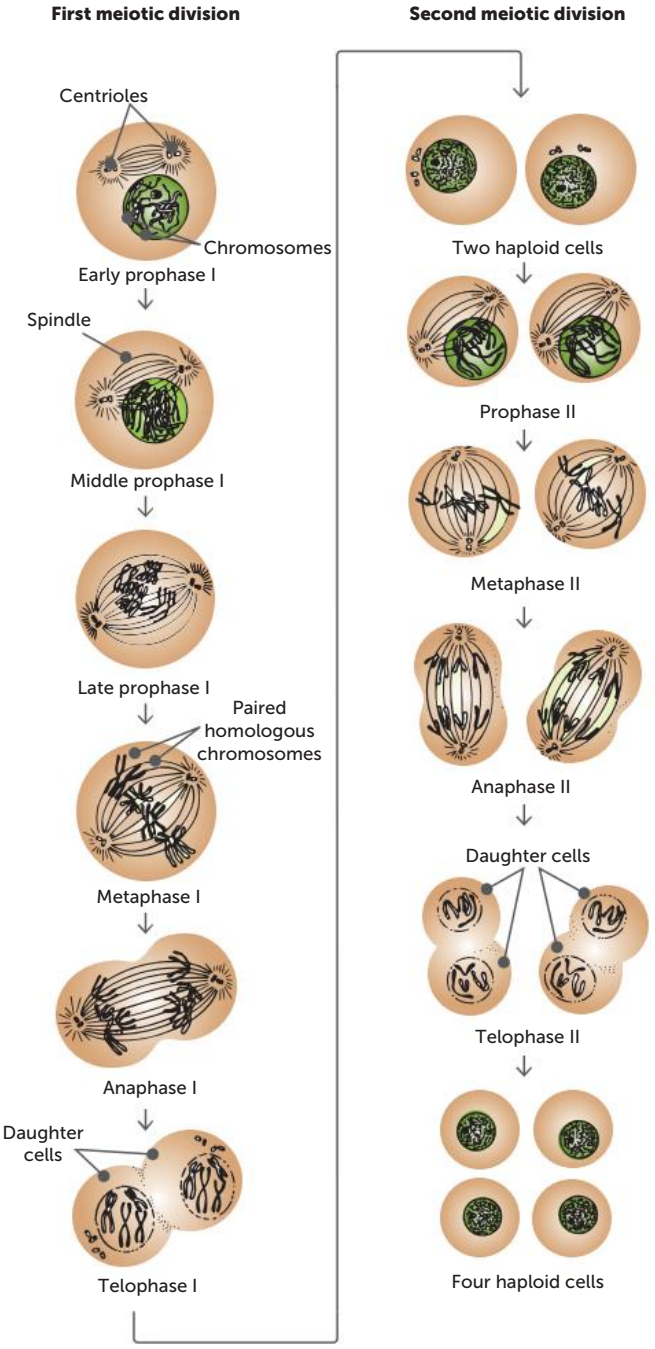


FIGURE 10.10 Stages of meiosis

While the chromosomes are shortening and thickening, a spindle forms, stretching between the poles of the cell. The paired chromosomes move towards the spindle fibres until, at **metaphase I**, they are arranged on the spindle fibres across the centre, or equator, of the cell. At **anaphase I**, the pairs of homologous chromosomes move apart, with *one* member of each *pair* (consisting of two chromatids) moving to each pole of the cell, resulting in 23 chromosomes moving to each pole of the cell. Thus, in the first division of meiosis, the number of chromosomes assembling at each pole of the cell is half the number present in the original cell. This is a major difference from the events occurring in mitosis.

During **telophase I**, the chromosomes decondense and the nuclear membranes may reform. Cytokinesis also occurs, with the cytoplasm dividing and the cell membrane forming to produce two cells.

Second meiotic division

During the second division, each daughter cell with its 23 chromosomes undergoes the same sequence of events, in which the chromatids separate and migrate to either end of the cell. This results in four haploid cells being formed.

During **prophase II**, a new spindle forms at each end of the original spindle and usually at right angles to the original. The chromosomes in each cell gradually move towards the equator, so that at **metaphase II** they are arranged on the new spindle. The centromeres then divide, so that each chromatid is now a separate chromosome. These new chromosomes migrate to opposite poles of the cell (**anaphase II**). Nuclear membranes begin to form and the cytoplasm starts to divide (**telophase II** and cytokinesis). By the end of the second division, four new cells have been formed, each with half the number of chromosomes of the original cell.

Key concept

The two stages of meiosis result in four haploid daughter cells.

Differences between mitosis and meiosis

The differences between mitosis and meiosis reflect their roles in the human body. Both processes involve the replication of DNA to produce a doubling of the number of chromosomes prior to cell division taking place. However, mitosis produces *diploid* cells for growth and repair within the tissues, whereas meiosis produces *haploid* gametes for sexual reproduction.

TABLE 10.3 Comparison of mitosis and meiosis

MITOSIS	MEIOSIS
One duplication of chromosomes and one nuclear division.	One duplication of chromosomes and two nuclear divisions.
Produces two diploid cells.	Produces four haploid cells.
Homologous chromosomes do not pair.	Homologous chromosomes pair up.
Chromatids separate so that each new cell gets a complete set of daughter chromosomes.	At first meiotic division, members of homologous pairs separate so that new cells get a haploid set of chromosomes. At second division, chromatids separate, giving four haploid cells.
Chromosomes do not change their genetic make-up.	Genetic make-up of chromosomes can be changed through crossing over.
Produces new cells for growth and repair.	Produces haploid gametes for sexual reproduction.

Questions 10.2

RECALL KNOWLEDGE

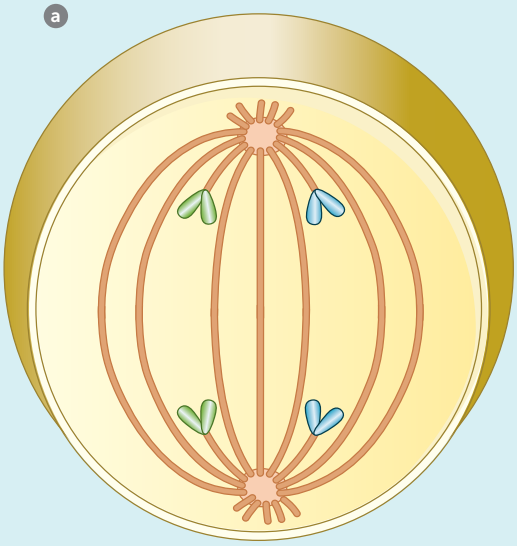
1 Where does meiosis occur?

2 Match the stage of meiosis with the process.

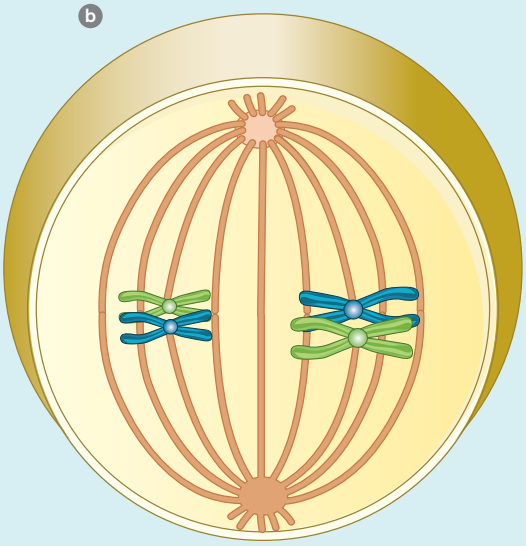
STAGE	PROCESS
Interphase	Homologous pairs separate
Meiosis I	DNA replicates
Meiosis II	Sister chromatids separate

3 For each of the diagrams below, name the phase of meiosis and state how you decided this.

a



b



4 How many chromosomes are in a cell at the
   a start of meiosis?
   b end of meiosis I?
   c end of meiosis II?

5 Define 'diploid'.

APPLY KNOWLEDGE

6 The Tasmanian devil has a diploid number of 14. State its haploid number.

7 Draw a Venn diagram to compare and contrast mitosis and meiosis.

8 Explain why meiosis has two stages, whereas mitosis has only one.

10.3 VARIATION IN DAUGHTER CELLS

One characteristic of meiosis is that it is able to produce daughter cells that vary in their genetic information. This is important, as it means that the offspring will differ from one another. Variation is a result of crossing over, non-disjunction and random (or independent) assortment.

Crossing over

An important feature of meiosis occurs during the prophase of the first meiotic division. When the homologous chromosomes are paired, the chromatids may cross, break and exchange segments. This is called **crossing over** and the point where two chromatids cross is called a **chiasma** (plural: **chiasmata**). Crossing over can result in a new combination of alleles along the chromosome. This is called **recombination**. Therefore, crossing over creates new combinations of genes so that the chromosomes passed on to the offspring are not exactly the same as those inherited from the parents.

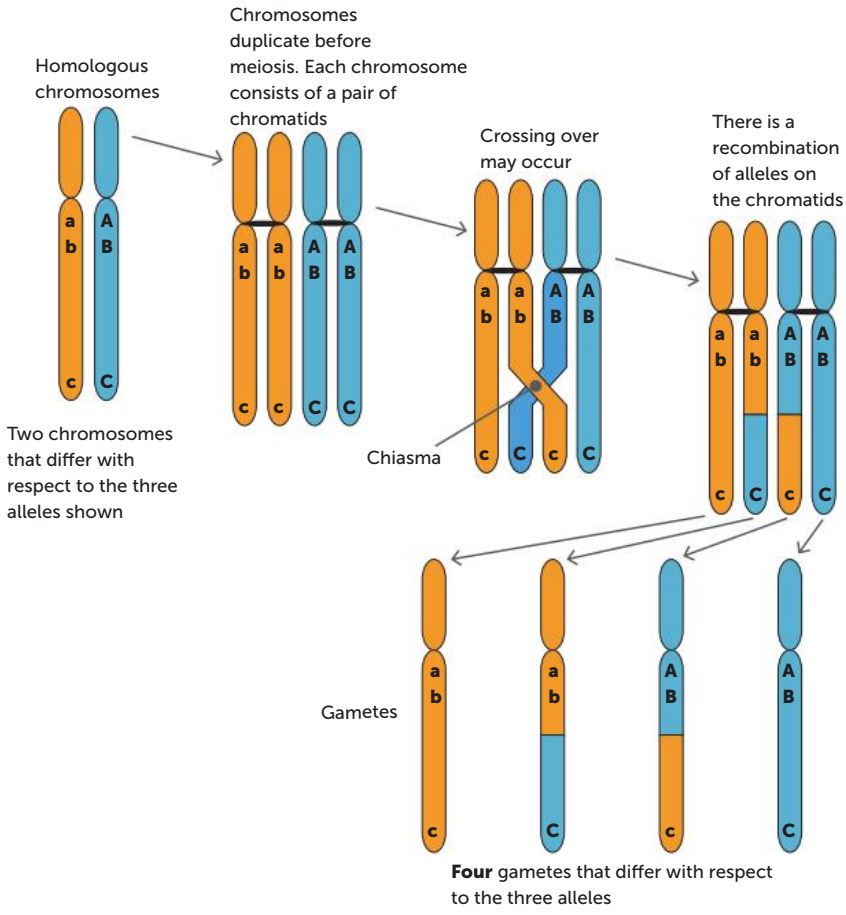


FIGURE 10.11 Crossing over and recombination

Non-disjunction

During the first division of meiosis, the homologous chromosomes pair and then separate. Sometimes one or more of the chromosome pairs may fail to separate when the cell divides. In the second meiotic division, one or more of the chromatids may fail to separate. These situations are called **non-disjunction**, and they result in one of the daughter cells receiving an extra chromosome and the other daughter cell lacking that chromosome.

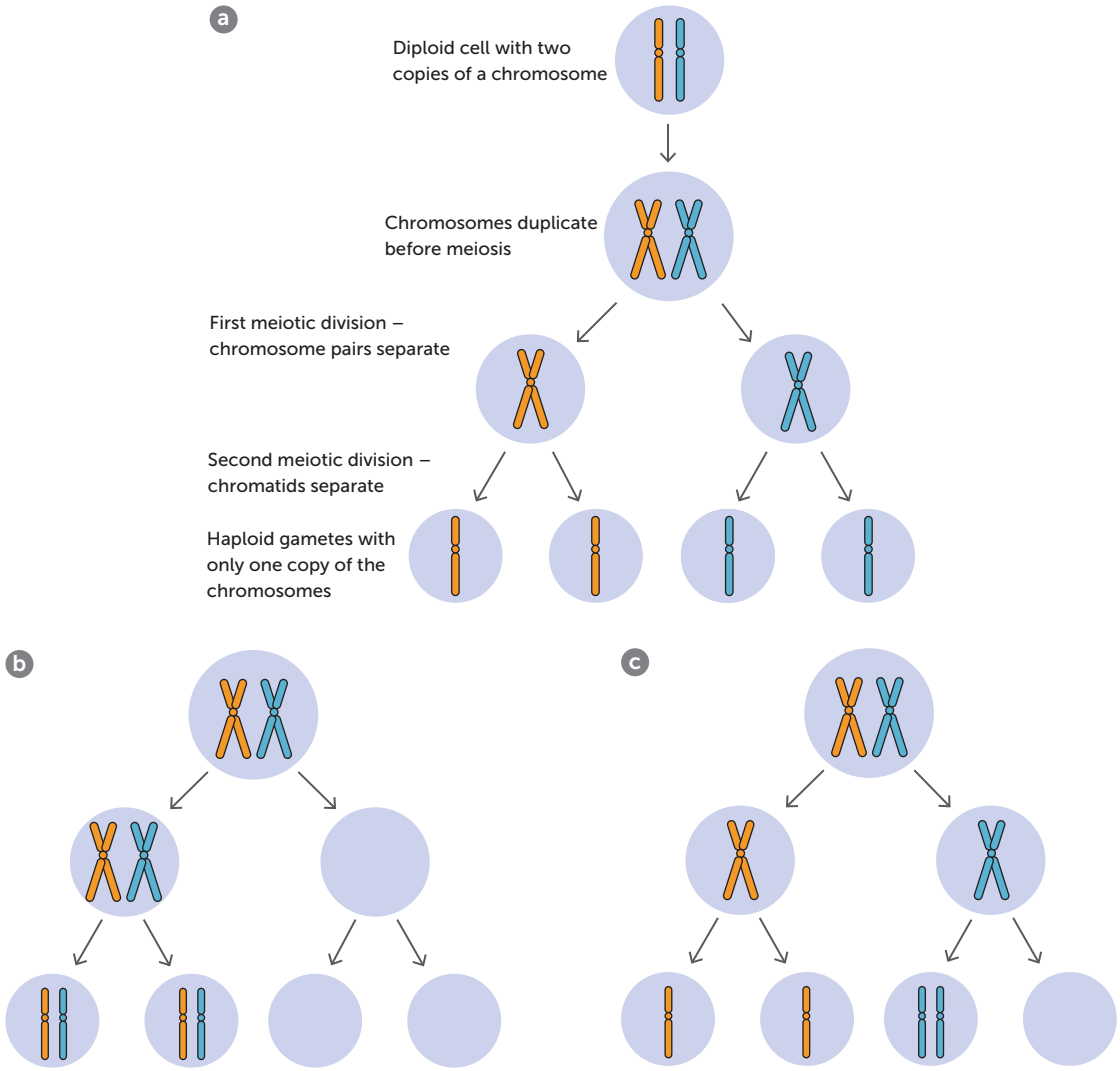
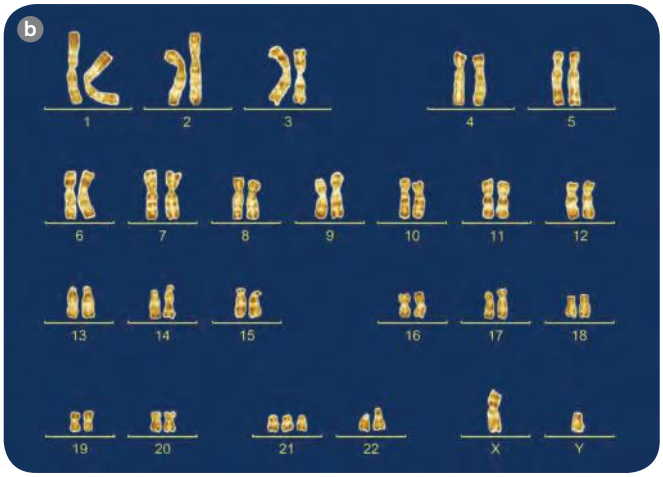


FIGURE 10.12
a Normal meiosis; **b** Non-disjunction in the first meiotic division; **c** Non-disjunction in the second meiotic division

In humans, if non-disjunction occurs in one of the chromosome pairs during meiosis, the resultant gametes will have either 24 chromosomes or 22 chromosomes instead of the normal 23. After fertilisation with a normal gamete from the opposite sex, the zygote produced will have either 47 or 45 chromosomes, respectively. This will produce quite unexpected characteristics in the offspring. Usually, such changes to the chromosome number cause severe and distinctive birth defects, and miscarriage often occurs early in the pregnancy.

Trisomy is a condition in which an individual inherits an extra copy of a chromosome – three copies instead of the normal two. One such chromosomal defect that occurs relatively frequently, especially in children of older mothers, is **Down syndrome**, or **trisomy 21**. In this disorder, an extra copy of chromosome 21 results in a characteristic facial appearance, variable degrees of intellectual disability and physical abnormalities.

Fairfax Photos/Angela Wylie



Science Photo Library/Pr. Philippe Vago, ISM

FIGURE 10.13 a A child with Down syndrome and her family; **b** A karyotype of Down syndrome (note the extra chromosome 21)

There are cases where trisomy occurs with other human chromosomes. An extra chromosome 13 (trisomy 13, or Patau syndrome) produces individuals with intellectual disability, a small head, extra fingers or toes, a cleft palate and/or cleft lip, and malformations of the ears and eyes. An extra chromosome 18 (trisomy 18, or Edwards syndrome) results in the individual having an intellectual disability and defects in the eyes, ears, hands and head.

Monosomy is where an individual is missing a chromosome – they have only one copy instead of the normal two. Like trisomy, monosomy usually results in severe malformations and often miscarriage.

Partial monosomy and partial trisomy can also occur. In **partial monosomy**, part of a chromosome is missing – part of the chromosome has two copies, but part has only one copy. **Partial trisomy** occurs when part of an extra chromosome is attached to one of the other chromosomes. Partial trisomy 21 can result in many of the symptoms of Down syndrome.

Random (or independent) assortment

During the first meiotic division, the homologous pairs of chromosomes separate. When these pairs of chromosomes separate, they do so at random. One member of each pair moves to one pole of the cell, while the other member of the pair moves to the opposite pole. An important feature of meiosis is that when the chromosomes move apart during the first meiotic division, they do so independently. The way one pair of chromosomes separates is unaffected by the way any of the other pairs separate. For example, the copy of chromosome 1 that an egg cell receives in no way influences which of the two possible copies of chromosome 5 it gets. This random, **independent assortment** takes place for each of the 23 pairs of human chromosomes. That means any single human egg receives one of two possible chromosomes 23 times. The total number of possible chromosome combinations is 2^{23}, which is approximately 8.4 million. And that is just for the eggs! The same **random assortment** goes on as each sperm cell is produced, too. Thus, when a sperm fertilises an egg, the resulting zygote contains a combination of genes arranged in an order that has probably never occurred before and will probably never occur again.

9780170449090

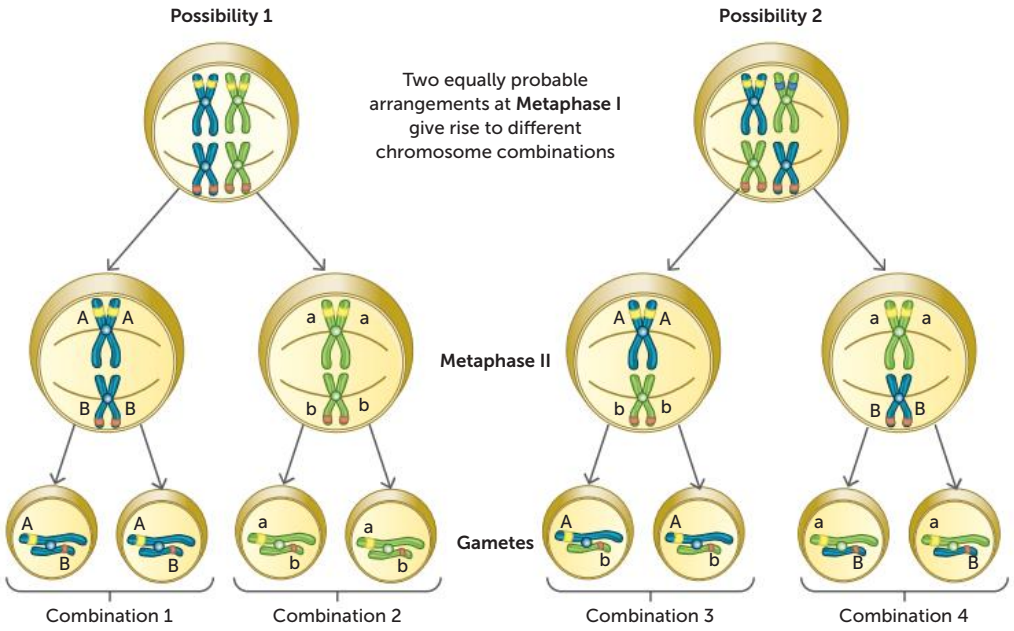


FIGURE 10.14 There are four possible combinations of chromosomes in gametes produced by a cell with just four chromosomes (two pairs)

Activity 10.3
Modelling meiosis

Key concept

Gametes, produced by meiosis from the same parent cells, differ due to crossing over, non-disjunction and random assortment.

Questions 10.3

RECALL KNOWLEDGE

1. List three processes that lead to variation between daughter cells.
2. Define 'chiasma'.
3. Explain how non-disjunction can lead to Down syndrome.
4. Draw a series of diagrams to show how non-disjunction in the first meiotic division can lead to trisomy.
5. Describe random (or independent) assortment with reference to chromosomes.

APPLY KNOWLEDGE

6. Explain the difference between crossing over and recombination.
7. If a cell has 15 pairs of chromosomes, how many different combinations of chromosomes are there in the gametes?

10.4 CANCER

Normally, the division of cells is controlled. However, under certain conditions this control is lost, and the cells divide uncontrollably. This condition is known as **cancer**. This uncontrolled growth of abnormal cells produces a mass, or **tumour**. As this process may occur in almost any type of tissue, there are many different types of cancer – for example, breast cancer, brain cancer, leukaemia, lung cancer and skin cancer.

Cancer cells do not differentiate into the normal tissue cells that surround the tumour. They can therefore be easily identified with a microscope. Some tumours are **malignant**, which means the tumour cells are able to spread to other parts of the body. This is known as **metastasis**. In this way, **secondary tumours** may develop in parts of the body well away from the original tumour.

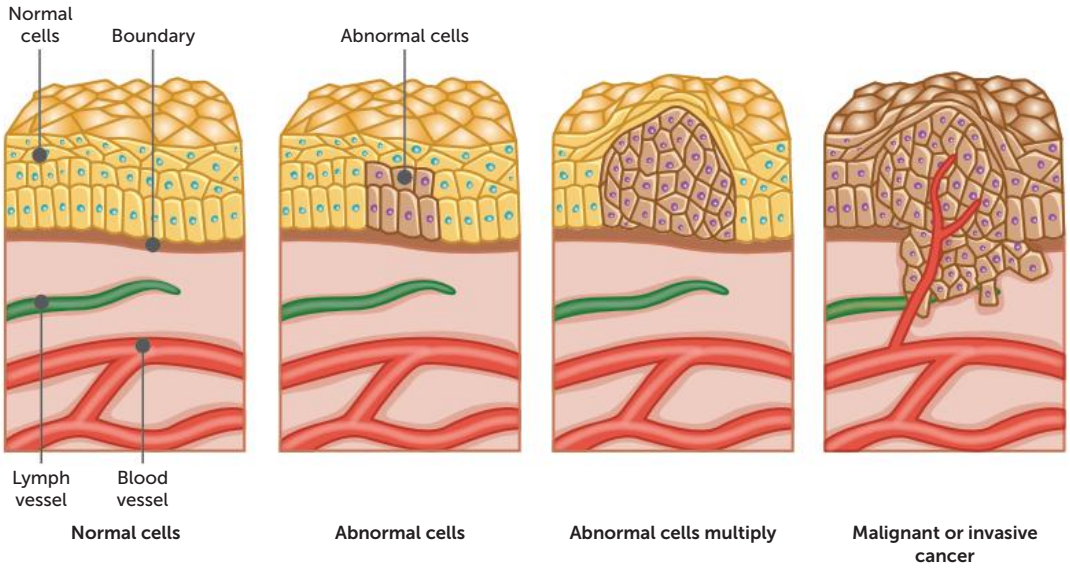
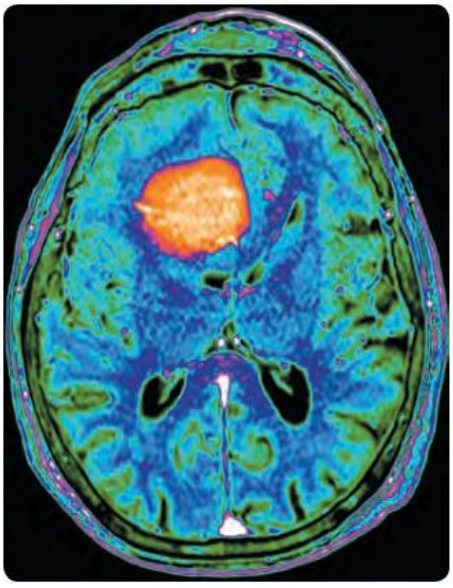


FIGURE 10.15 The process of cancer forming a tumour

Some tumours are not malignant as they are not able to invade normal tissues, blood or lymph vessels, and so do not spread to other parts of the body. These tumours are called **benign**. Benign tumours grow and press on surrounding tissues. Such tumours can be dangerous if they exert pressure on vital organs such as the brain. However, because a capsule often surrounds them, they are usually easily removed.



Science Photo Library/Zephyr

FIGURE 10.16 A tumour is an abnormal mass of cells that can affect the tissue around it – for example, the brain tumour visible on this MRI

Medical online: cancer information
Cancer Council
Cancer Council WA
Cancer Council NSW

Key concept

Cancer is a collection of cells that undergo uncontrolled cell division. This can result in the formation of masses of cells called tumours.

Causes of cancer

While the cause of some cancers is unknown, we do know that certain environmental factors called **carcinogens** can trigger malignant tumours. Cancer usually occurs only after long exposure to a carcinogen, and the cancer may develop many years after the exposure has ended. Other factors, such as genetics, can also affect the likelihood of developing cancer.

Carcinogens

A great many substances and forms of radiation have been found to be associated with cancers.

- Ultraviolet (UV) radiation, which is a part of sunlight, produces cancer of the skin, especially in people with light-coloured skin. Sunburn and overexposure to UV radiation are the main causes of skin cancer.
- X-rays are known to cause cancer. In Australia, exposure to X-rays is limited and controlled. The amount of radiation produced by modern machines poses little risk to patients from routine medical use.
- Ionising radiation, such as that produced by radium and ores of uranium, can cause cancer. A single exposure to a high dose may result in leukaemia. Radiation from the atomic bombs dropped on Hiroshima and Nagasaki in Japan at the end of World War II caused a significant increase in the incidence of cancers in the people of those cities.
- Viruses have been found to cause some forms of cancer. For example, the human papilloma virus (HPV) causes cancer of the cervix in women. A vaccine called Gardasil® that protects young women against some forms of HPV was introduced in Australia in 2007.
- Chemical carcinogens are widespread in modern society, but simple precautions can usually be taken to avoid excessive exposure. Some known chemical carcinogens are alcohol (excessive consumption), asbestos, soot and tar, organic solvents in glues and paints, and tobacco tar.

Prevention of cancer

Many cancers are associated with lifestyle factors, such as exposure to UV radiation, smoking, alcohol consumption and diet.

In Australia, the incidence of cancer has been reduced in two ways.

1. *By education:* The public has been made aware, through advertising and other education programs, of the need to limit exposure to carcinogens. An example was the very successful 'Slip! Slop! Slap!' program, introduced in 1981, to make people aware of the need to limit exposure of the skin to UV radiation. This message has now been expanded to 'Slip! Slop! Slap! Seek! Slide!'.
2. *By legislation:* Australian governments have passed laws to control exposure to carcinogens. For example, smoking is banned in most public places, advertising of tobacco is not permitted, and cigarettes must be sold in plain packaging with graphic health warnings. Standards have been imposed for the manufacture and operation of X-ray machines, and products containing asbestos have been banned. These and other measures have helped to reduce the incidence of cancer, but each of us still has a responsibility to minimise our own risks as far as possible.

Cancer Council Victoria

FIGURE 10.17 The original 'Slip, Slop, Slap' campaign effectively educated the public about the importance of protection against UV rays

9780170449090

Some positive steps that you can take to reduce the risk of cancer later in life are as follows:

- Avoid smoking.
- Use sunscreen, sunglasses, long-sleeved clothing, shade and hats to reduce exposure to UV radiation.
- If possible, stay out of direct sunlight between 10 a.m. and 3 p.m.
- Ensure that your diet has adequate fibre.
- Avoid being overweight or obese.
- Limit alcohol intake, if you choose to drink.
- Use protective clothing and a face mask when handling chemicals such as organic solvents or vinyl chlorides.

Early detection of cancer

Cancer is a leading cause of death in Australia. One in every two Australian men and one in three women will be diagnosed with cancer by the age of 85. Early detection is critical for the successful treatment of the disease. Tests are now available for a number of common cancers so that treatment may begin at a very early stage of tumour growth.

Cervical cancer

Any woman who has had sexual intercourse at any time in her life is at risk of developing cancer of the cervix. Cervical cancer is caused by the human papilloma virus (HPV), which is transmitted by genital skin contact during intercourse. In most cases, an infection clears up naturally in about 8 to 14 months, so most people are infected with HPV at some time in their lives and will never know it. In a small number of women, the infection does not clear up and abnormalities of cervical cells can develop.

In 1928, Dr George Papanicolaou, a Greek-born doctor working in the United States, discovered that changes occur in cervical cells before they become cancerous. A simple test for the presence of these abnormal cells, the Papanicolaou, or 'Pap', test was developed in which cells are collected from the cervix and smeared on to a microscope slide. The cells are then examined for abnormalities. This test, now known as the Cervical Screening Test, does not diagnose cancer; it detects early changes in cervical cells that may develop into cancer. Early treatment of the cancer is then possible.

A Cervical Screening Test every three to five years can prevent up to 90% of the most common form of cervical cancer, and so it is now one of the most preventable and curable of all cancers.

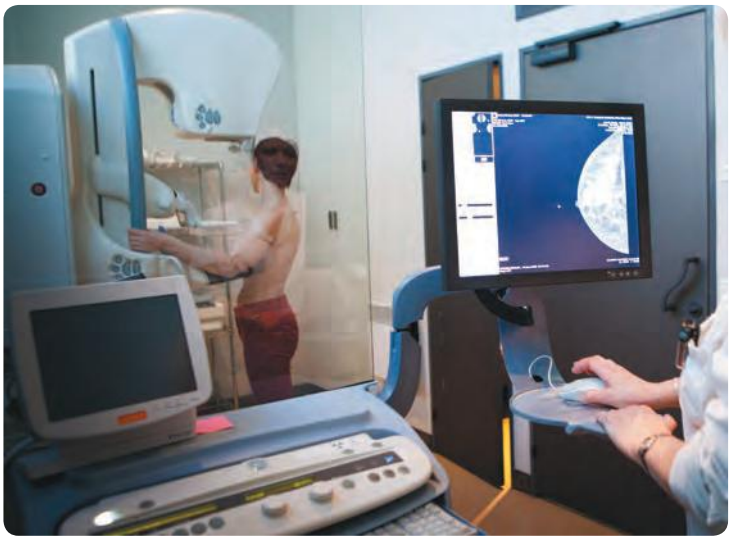
Breast cancer

Breast cancer is the most common type of cancer in Australian women and the second most common cause of cancer-related death. Since 1991, BreastScreen Australia has run a free screening program for Australian women aged 50 to 74, although those aged 40 to 49 and 75 or older are also able to take part.

TABLE 10.4 Most common causes of cancer-related deaths in Australia

WOMEN	MEN
1 Lung	1 Lung
2 Breast	2 Prostate
3 Bowel (colorectal)	3 Bowel (colorectal)

Screening is done by mammography, an X-ray of the breasts (Figure 10.18). The X-ray results in a mammogram, an X-ray picture on which tumours as small as about 1 cm in diameter can be detected. Digital mammography uses a computer instead of X-ray film to record the images of the breast.



Science Photo Library/BSIP/Emelie-Benoist

FIGURE 10.18 Digital mammography

Bowel cancer

Bowel cancer, or colorectal cancer, is a malignant tumour that develops in the large intestine – the colon or the rectum. It can be treated successfully if diagnosed early, but there are often no symptoms and at present fewer than 40% of bowel cancers are detected in the early stages.

Australians between 50 and 74 years of age are invited to take part in a bowel cancer screening program. Eligible persons are sent a simple test called a faecal occult blood test (FOBT) every two years. The test, for blood in the faeces, is done at home and then mailed to a laboratory for analysis. Very small amounts of blood, not visible to the naked eye, can be detected.

Blood in the faeces can come from polyps or from bowel cancer. Polyps are small growths inside the colon or rectum. Most bowel cancers develop from polyps, although not all polyps become cancerous. However, removal of polyps reduces the risk of bowel cancer.

If the FOBT test is positive, patients are usually referred for a colonoscopy, a visual examination of the inside of the large intestine using an instrument called a colonoscope.

Prostate cancer

Unlike cancers of the cervix, breast or bowel, there is no screening program in Australia for prostate cancer. Trials in the United States and in Europe showed little benefit in such screening. Many prostate cancers grow very slowly and do not require any treatment, but other forms are life-threatening because they grow and spread rapidly. Unfortunately, there is no test that distinguishes between these cancers.

Aggressive prostate cancer can be cured if diagnosed while it is still confined to the prostate gland. There are three diagnostic methods: digital rectal examination (DRE), prostate-specific antigen (PSA) blood test, and biopsy.

In DRE, the doctor inserts a gloved finger into the anus, from where it is possible to feel part of the surface of the prostate gland. Any swelling, hardening or irregularities of the surface may indicate cancer. The problem is that only part of the prostate surface can be felt, so some irregularities may be beyond reach.

The PSA test checks the blood for the presence of a particular protein produced by the prostate gland. If the PSA is rising, it may indicate the presence of a prostate tumour.

If rectal examination or a PSA test indicates the possibility of cancer, then a biopsy can be performed. A biopsy is a small sample of tissue that can be checked for cancer cells. In the case of the prostate, a spring-loaded needle is used and several samples are taken. The procedure is often done under general anaesthetic. Tissue samples can be examined to determine the presence of tumour cells and, if they are present, the type of tumour. A decision can then be made about treatment.

Unfortunately, all these diagnostic procedures have limitations and side effects, and so experts disagree on the best way to tackle prostate cancer. Because many prostate cancers

develop so slowly that a man may die *with* the disease rather than *of* the disease, some doctors say it is better for men not to know whether they have prostate cancer. The peak Australian body for prostate cancer, the Prostate Cancer Foundation of Australia, strongly rejects this point of view.

Individual responsibility

Each of us should be familiar with our own body and should see a doctor if any suspicious change is noticed. For example, a breast lump, a lump in a testicle, a change in a mole, a change in bowel or bladder habits, any sore that does not heal, persistent cough or hoarseness, or indigestion or difficulty swallowing could be an indication of cancer.

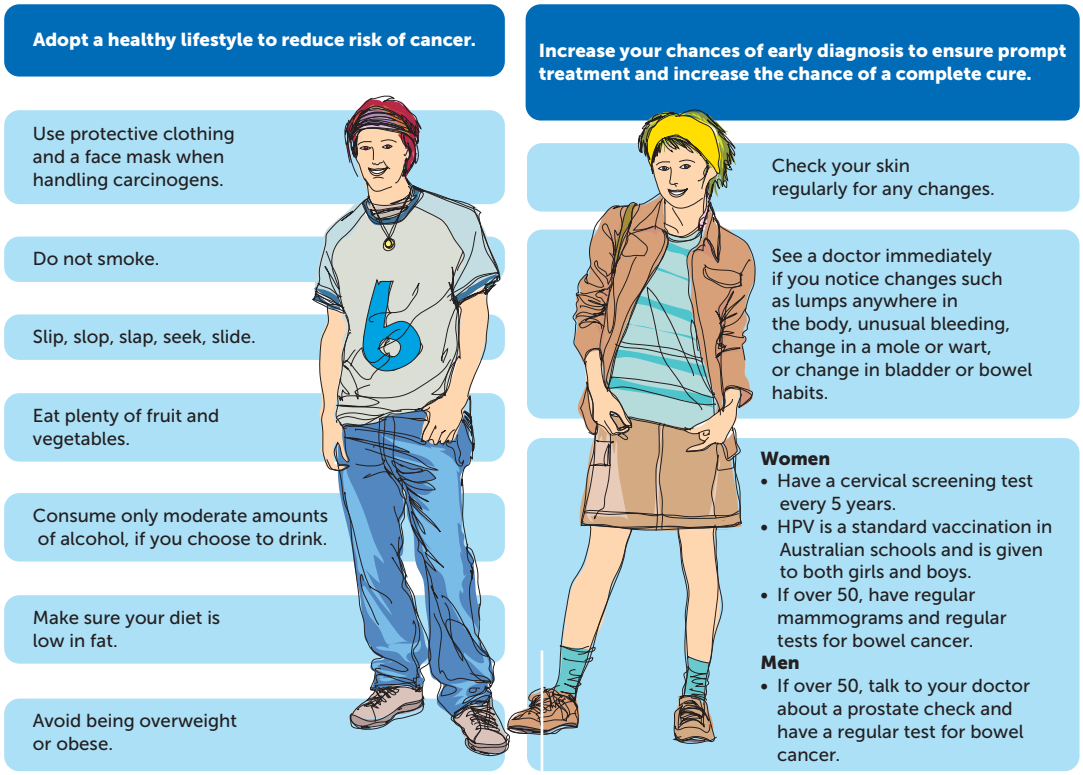


FIGURE 10.19 Take responsibility for your own health: reduce your risk of getting cancer later in life and increase the chance of early diagnosis so that any cancer can be treated

Activity 10.4
Investigating the incidence of cancer in Australia

Questions 10.4

RECALL KNOWLEDGE

1. Define 'cancer'.
2. List five different types of cancer.
3. List three carcinogens.
4. Describe how cervical cancer can be detected in its early stage.
5. Name the diagnostic tests for prostate cancer.

APPLY KNOWLEDGE

6. Explain how cancer cells are different from normal cells.
7. Explain why tumours can be identified when a sample is viewed under a microscope.
8. Compare and contrast benign tumours and malignant tumours.
9. Explain how education has been able to reduce the incidence of some cancers.
10. Suggest why some people who receive the faecal occult blood test kit still do not do the test.

CHAPTER 10 ACTIVITIES

ACTIVITY 10.1 Modelling mitosis and cytokinesis

In science, a model is a simplified version of a complex process. This helps us to understand scientific concepts and processes.

Your task

In groups of approximately 10, role-play mitosis and cytokinesis. Take a video or a series of photos of the model. Add labels or a voice-over to explain what is happening in each stage.

ACTIVITY 10.2 Observing mitosis

You can observe the phases of mitosis by looking at dividing cells under a microscope. Your teacher will provide prepared slides of dividing cells. Often these will be cells from the root tip of an onion. Mitosis in plant and animal cells follows the same sequence of events, but mitosis is often easier to observe in plant cells.

What to do

Use a microscope to observe cells undergoing mitosis. Look for cells that are in prophase, metaphase, anaphase and telophase. If you have forgotten how to use a microscope correctly, refer to Activity 2.1 in Chapter 2.

Results

1. Draw a cell that is in each of the four phases.
2. You cannot see the spindle in any of the cells. Suggest why it cannot be seen.
3. Estimate the number of chromosomes in the cells you are observing. How does your estimate compare with that of others in your class?
4. If you observed onion cells, what major difference did you see between those cells and the animal cells we have discussed in this chapter?

ACTIVITY 10.3 Modelling meiosis

You will need

For each pair: a large sheet of paper, laminated board or benchtop to write on; eight pipe-cleaners (four each of two different colours); wire ties; pencil or whiteboard marker; eraser

What to do

1. Create a model of a cell with six chromosomes of different colours (three homologous pairs), showing the cell membrane, nuclear membrane, chromosomes and centrioles. Some ideas are:
   - Draw the cell on paper or a white board.
   - Use pipe-cleaners to make a cell.
   - Use plasticine to make a cell.
   - Use lollies to make a cell.
   - Draw your cell using a program such as Keynote or PowerPoint.
   - Make a virtual cell.
2. Take a photo of your cell.
3. Take your cell through the process of meiosis, starting with the replication of DNA during interphase then meiosis I, cytokinesis I, meiosis II and cytokinesis II. At each stage, take a photo to show.

4 Use your photos to create a stop motion video.
5 Add labels to identify each stage of meiosis.

Extension: include crossing over or non-disjunction during the appropriate stages.

Studying your results

1 What is meant by a 'model' in science?
2 With respect to the colours of the chromosomes, how many different types of gametes did your model produce? How many colour combinations are possible?
3 Suppose the chromosome number of your cell was 10. How many combinations of chromosomes would now be possible?
4 Humans have a chromosome number of 46. What can you say about the number of possible chromosome combinations in human eggs and sperm?
5 Why is it that children of the same parents do not inherit identical chromosomes (except for identical twins)?
6 How did your movie demonstrate independent assortment of chromosomes?

ACTIVITY 10.4 Investigating the incidence of cancer in Australia

Many people are treated successfully for cancer each year, but cancer is still a major cause of death in Australia. Use references to find out:

- which cancers are most common in Australia
- whether there is any relationship between the type of cancer and where people live in Australia
- the age groups at which particular cancers are more common in Australia
- whether there are any upward or downward trends in the incidence of particular cancers in Australia.

You may use the weblinks for the Cancer Councils on page 262. There are also websites for organisations that deal with specific types of cancer. As with any information that you use from the Internet, make sure that it has come from a reliable source.

9780170449090

CHAPTER 10 SUMMARY

- Cells reproduce to produce new cells and replace old, dead or damaged cells.
- The cell cycle is made up of the first growth phase, synthesis phase, second growth phase and mitotic phase.
- Mitosis, including DNA replication, ensures that the new cells are an exact copy of the parent cell.
- The stages of mitosis are interphase, prophase, metaphase, anaphase and telophase.
- Cytokinesis, or the division of the cytoplasm, occurs simultaneously with telophase.
- Stem cells are undifferentiated cells that are able to form either new stem cells or specialised cells.
- Stem cells can be classified as totipotent, pluripotent or multipotent.
- Gametes, produced by meiosis, have half the number of chromosomes as somatic cells.
- During the first stage of meiosis the homologous chromosomes separate, while during the second stage the chromatids separate.
- Both stages of meiosis consist of prophase, metaphase, anaphase and telophase.
- The daughter cells of meiosis vary in their genetic information due to crossing over, non-disjunction and random assortment.
- Crossing over can occur during the first stage of meiosis when homologous chromosomes exchange segments.
- Non-disjunction may occur during the first stage of meiosis if the homologous chromosomes do not separate, or during the second stage of meiosis if the chromatids do not separate.
- Random, or independent, assortment is the homologous chromosomes separating in a manner independent of one another. This results in the combination of chromosomes from each homologous pair varying between daughter cells.
- Cancer is the uncontrolled division of cells, producing masses called tumours.
- Malignant tumours may spread through the body, while benign tumours do not.
- Carcinogens are factors that may trigger malignant tumours. These include UV radiation, X-rays, ionising radiation, viruses, and some chemicals such as tobacco and alcohol.
- Early detection of cancer increases the chance of survival. Cervical cancer may be detected by a Cervical Screening Test; breast cancer can be detected through a mammogram; bowel cancer is detected by a faecal occult blood test; and prostate cancer is detected by a rectal exam, blood test and biopsy.

9780170449090

CHAPTER 10 GLOSSARY

Anaphase The third phase of mitosis, during which the daughter chromosomes are drawn to opposite ends of the cell

Anaphase I The third phase of the first stage of meiosis, during which the homologous chromosomes move to opposite ends of the cell

Anaphase II The third phase of the second stage of meiosis, during which the daughter chromosomes move towards opposite ends of the cell

Benign Not able to spread to other parts of the body

Cancer A malignant growth; one that has the capability of spreading to other body parts

Carcinogen A cancer-causing agent

Cell cycle The events that take place from one cell division to the next

Chiasma The point at which crossing over occurs between chromatids; plural: chiasmata

Crossing over The interchange of the parts of the chromatids of a homologous pair of chromosomes during the first stage of meiosis; it creates new combinations of alleles

Cytokinesis The division of the cytoplasm to form two daughter cells

Differentiation of cells The process by which unspecialised cells develop special characteristics to suit particular functions

Diploid number The number of chromosomes in a cell with both chromosomes from each homologous pair

Down syndrome *see* trisomy 21

Fertilisation Fusion of sperm and egg

G_0 phase The phase of the cell cycle when the cell is functioning but not preparing for division

G_1 phase The first growth phase of the cell cycle

G_2 phase The second growth phase of the cell cycle

Gamete A sperm or egg cell

Haploid number The number of chromosomes in a cell with only one chromosome from each homologous pair; half the diploid number

Homologous chromosomes The pairs of chromosomes containing genes that control the same characteristics

Independent assortment The random combination of alleles due to allele pairs separating independently of one another

Interphase The stage in the life cycle of a cell when it is not dividing; the stage between mitotic divisions

M phase The phase of the cell cycle when the cell divides into daughter cells; also known as the mitotic phase

Malignant Able to spread to other parts of the body

Meiosis A type of cell division resulting in sperm or eggs; the sperm or eggs have half the chromosome number of the parent cell

Metaphase The second phase of mitosis, during which the chromosomes (pairs of chromatids) line up across the centre of the cell

Metaphase I The second phase of the first stage of meiosis, during which the homologous chromosomes line up on the equator

Metaphase II The second phase of the second stage of meiosis, during which the chromosomes line up on the equator

Metastasis The spreading of tumour cells to form secondary tumours in different parts of the body

Mitosis The process of division of the nucleus of a cell in which the two daughter nuclei have the same number and type of chromosomes as the parent nucleus; often used loosely to mean cell division

Monosomy Where an individual has only one copy of a chromosome instead of two

Multipotent stem cells Stem cells that are able to give rise to a limited number of other

9780170449090

cell types; for example, blood stem cells will give rise to red blood cells, white blood cells and platelets

Non-disjunction When one or more of the chromosome pairs fail to separate during meiosis

Partial monosomy When a person has part of a chromosome missing

Partial trisomy When a person has part of an extra chromosome

Pluripotent stem cells Stem cells that are able to give rise to many, but not all, of the cell types necessary for foetal development

Prophase The first phase of mitosis, during which the chromosomes become visible, the nuclear membrane breaks down and the spindle forms

Prophase I The first phase of the first stage of meiosis, during which the chromosomes become visible, the nuclear membrane breaks down and the spindles form

Prophase II The first phase of the second stage of meiosis, during which the nuclear membrane disappears and the spindles form

Random assortment *see* independent assortment

Recombination A changing of the order of alleles along a chromosome

S phase The synthesis phase of the cell cycle, when DNA molecules duplicate

Secondary tumour Cancer that has spread from the original cancer

Stem cell Cell that has the ability to produce different types of body cells

Telophase The final phase of mitosis, during which the daughter chromosomes group at opposite ends of the cell and two daughter nuclei form

Telophase I The fourth phase of the first stage of meiosis, during which the new nuclear membrane forms

Telophase II The fourth phase of the second stage of meiosis, during which the new nuclear membrane forms

Totipotent stem cells Stem cells that are able to create any of the types of cell necessary for embryonic development

Trisomy A condition in which an individual inherits an extra copy of a chromosome

Trisomy 21 A genetic disorder resulting from an extra copy of chromosome 21; affected individuals have an altered physical appearance and variable degrees of intellectual disability; also called Down syndrome

Tumour An abnormal mass of tissue resulting from uncontrolled division of cells

9780170449090

CHAPTER 10 REVIEW QUESTIONS

Recall

1 Describe the function of the DNA in a cell.

2 a What is the cell cycle?
b Describe what happens in the four phases of the cell cycle.

3 Name three places where mitosis would be occurring in the body of a healthy adult human.

4 Define 'carcinogen' and list five examples.

5 Describe the most common tests for:
a bowel cancer
b breast cancer
c prostate cancer
d cervical cancer.

Explain

6 Explain the difference between a chromatid and a chromosome.

7 Explain why cell reproduction is necessary in the places listed in Question 3.

8 Explain how mitosis ensures that each daughter cell has exactly the same genetic information as the parent cell.

9 Explain how each of the following lead to variation in daughter cells:
a independent assortment
b non-disjunction
c crossing over.

10 Explain the difference between a benign and a malignant tumour.

Apply

11 a Use a series of diagrams, or a written description, to show the events that take place during meiosis.
b Explain why meiosis is essential in sexually reproducing organisms.
c Explain the difference between haploid and diploid cells.

12 The genes for hair colour and eye colour are located close to each other on the same chromosome. Explain why these traits are usually inherited together.

13 Skeletal muscle cells and most nerve cells remain in the G_0 phase of the cell cycle. Is it likely that these cells would be dividing? Explain your answer.

14 Explain the main differences between the processes of mitosis and meiosis. Relate the differences to the type of cells produced by each process.

15 How many chromosomes are present in a cell in a human ovary during each of the following stages of meiosis?
a Prophase of the first meiotic division
b At the end of telophase of the first division
c Prophase of the second meiotic division
d At the end of telophase of the second division

16 Variation only occurs when organisms reproduce sexually. When a single-celled organism such as an amoeba reproduces asexually, the two new amoebae are identical to the parent. Explain why asexual reproduction does not produce variation.

17 Use a table to summarise the advantages and disadvantages of the different diagnostic tools for prostate cancer.

Extend

18 What do you think would happen if the spindle fibres did not form in a cell that was undergoing mitosis?

19 Explain why medical scientists hope that many diseases that have so far been untreatable may be able to be treated using stem cells.

9780170449090

20 The frequency of non-disjunction of chromosome 21 (Down syndrome) increases with the age of the mother. Find out how age is thought to contribute to non-disjunction.

21 **a** List as many reasons as you can for the fact that Australia has the highest incidence of skin cancer in the world.

b How can you change your habits to reduce the risk of skin cancer?

c Describe any recommended changes in beach wear that are aimed at reducing exposure to UV radiation.

22 List reasons why our exposure to carcinogens is greater today than it has been in the past.

9780170449090

11 THE STRUCTURE OF THE REPRODUCTIVE SYSTEMS ALLOWS REPRODUCTION

UNIT 2 CONTENT

SCIENCE INQUIRY SKILLS

» design investigations, including the procedure(s) to be followed, the materials required, and the type and amount of primary and/or secondary data to be collected; conduct risk assessments; and consider research ethics, including animal ethics

SCIENCE UNDERSTANDING

Human reproduction

» the production of offspring is facilitated by the structure and function of the male and female reproductive systems in producing and delivering gametes for fertilisation and providing for the developing embryo and foetus

» both male and female reproductive systems are regulated by hormones, including the regulation of the menstrual and ovarian cycles

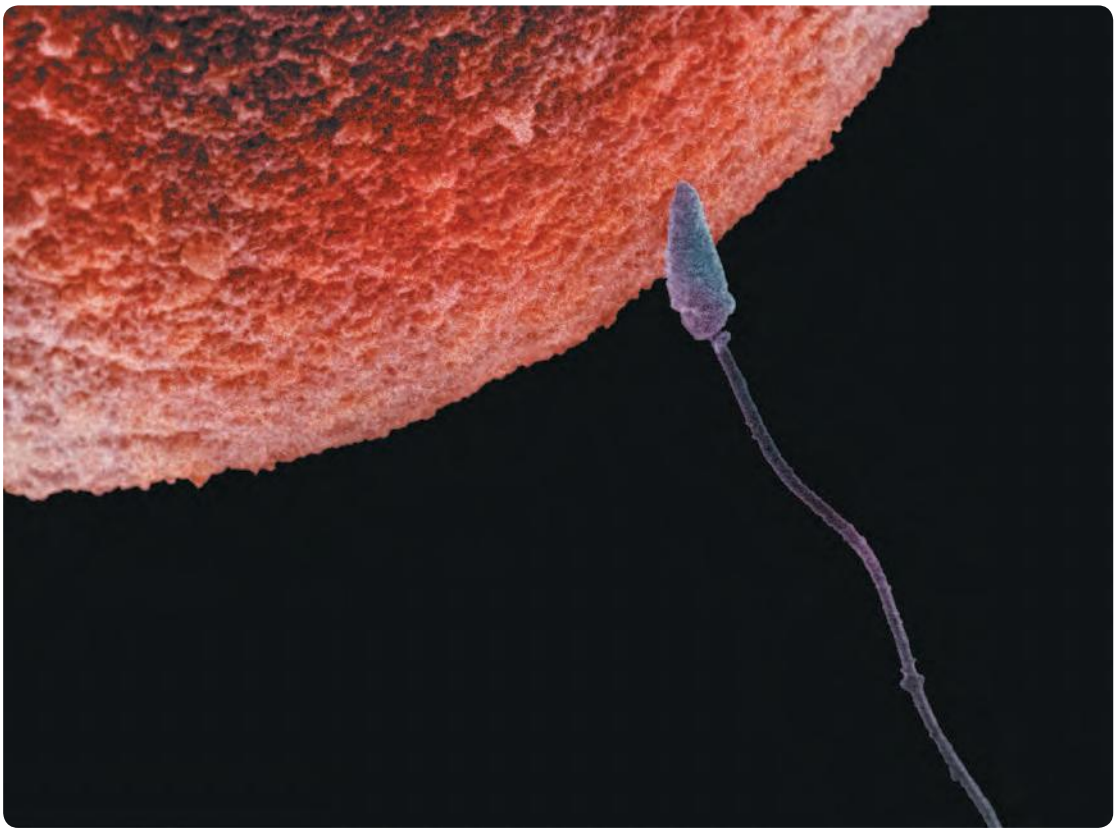
» human gametes are produced through spermatogenesis and oogenesis, which are specific forms of meiosis, but varying significantly in process and products

Source: School Curriculum and Standards Authority, Government of Western Australia

9780170449090

The survival of any species depends on reproduction, and the human species is no exception. To ensure the continuation of our species, some members must produce new individuals to replace those who inevitably die.

Humans reproduce sexually – a process involving the joining of male and female sex cells to produce a single cell, called a **zygote**, that develops to produce a new individual.



Science Source/Don W Fawcett

FIGURE 11.1 Male gamete (sperm) and female gamete (egg)

The gonads are the **primary sex organs**; they produce the gametes. Other organs are essential for reproduction; they store the gametes, bring them together for fertilisation and support the developing baby. These are the **secondary sex organs**.

11.1 STRUCTURE OF THE REPRODUCTIVE SYSTEMS

The reproductive system of humans is different from the other systems of the body because the organs making up the system in the male are quite unlike those of the female.

Male reproductive system

The male gonads consist of two testicles or **testes**, where the male gametes, **spermatozoa** (or **sperm**), are produced. The testes are held and supported in a skin-covered pouch called the **scrotum**. The scrotum appears to be a single pouch of skin, but internally it is divided into two sacs, each containing a single testis.

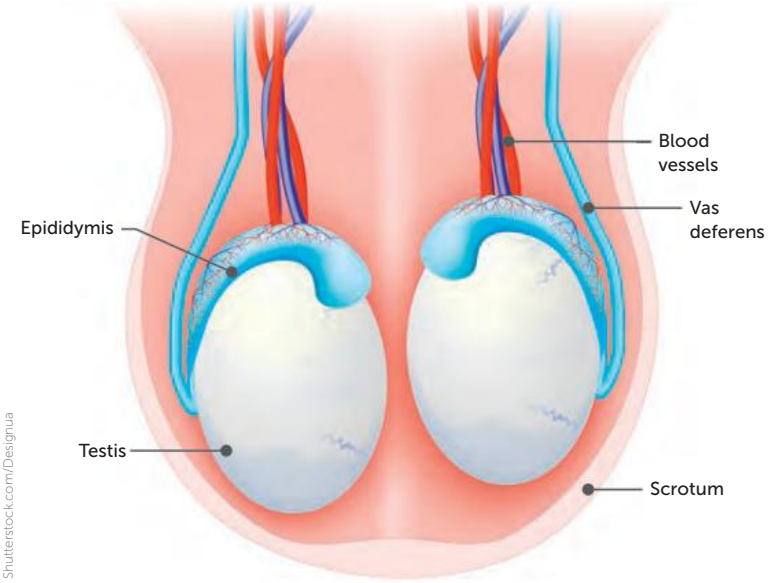


FIGURE 11.2 The testes are located in the two sacs of the scrotum

The production and development of sperm requires a temperature that is about 2°C lower than the normal body temperature. Therefore, the testes lie in the scrotum, outside the body cavity. With exposure to cold, contraction of smooth muscle fibres in the wall of the scrotum moves the testes closer to the body, where the temperature is slightly higher. If necessary, the same muscles can relax, moving the testes away from the body to keep them cooler.

Each testis is oval in shape, approximately 4.5 cm long, 2.5 cm wide and 3 cm thick. Internally they are divided into between 200 and 300 **lobules**, or compartments, filled with fine tubes called **seminiferous tubules**. The tubules are lined with cells that produce the male gametes. The seminiferous tubules in each compartment of the testis join to form a short, straight tubule. These straight tubules eventually join into ducts, which leave the testis and enter a structure called the **epididymis**. Between the seminiferous tubules are clusters of **interstitial cells** that secrete the male hormone testosterone.

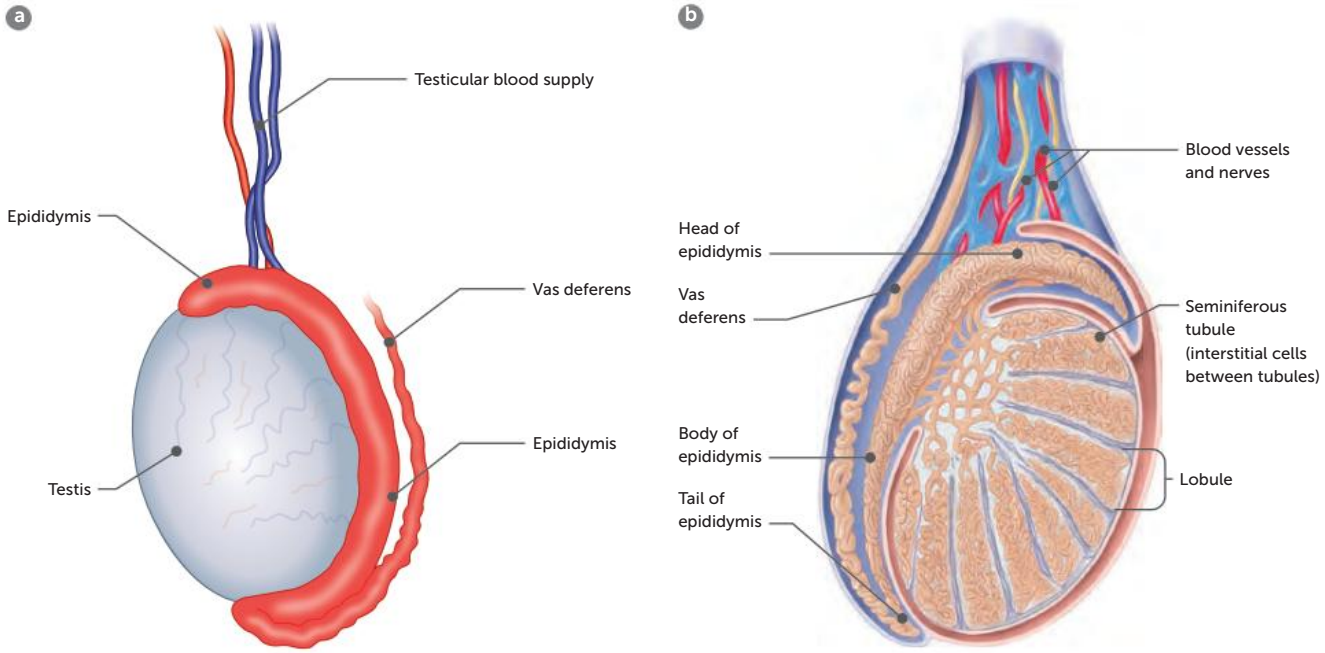


FIGURE 11.3 a External structure of the testis; **b** Cross-section through a testis and epididymis, showing the system of tubules

The epididymis is a highly folded tubule that fits against the rear surface of each testis. Sperm from the testis enter the tubule of the epididymis where they are stored for up to a month while they mature. If the tubule were unravelled, it would be about 5–6 m in length, allowing plenty of space for the storage of sperm.

The tubule of the epididymis continues to become the **vas deferens** (or **sperm duct**), which carries the sperm away from the testis. The vas deferens extends upwards from the testis, passes into the abdominal cavity and crosses the upper surface of the bladder. It then turns downwards, looping behind the bladder.

Under the bladder, the two vasa deferentia (plural form), one from each testis, join the **urethra,** which runs from the bladder, through the penis to the exterior. Therefore, the urethra is a duct for transporting both urine and sperm.

For transfer into a female's body, and to reach the egg for fertilisation, the sperm must be in a liquid. This liquid, the **semen** (or **seminal fluid**), nourishes and aids the transport of sperm. It is a mixture of secretions from three glands – the seminal vesicles, the prostate gland and the bulbo-urethral gland.

- The **seminal vesicles** are a pair of pouch-like organs about 5 cm in length located behind the urinary bladder. They secrete a thick fluid that is rich in sugars and makes up about 60% of the volume of semen.
- The **prostate gland** is where the two vasa deferentia join the urethra. It is a single gland, shaped like a doughnut, which surrounds the urethra just below the bladder. It secretes a thin, milky, alkaline fluid that also becomes part of the semen.

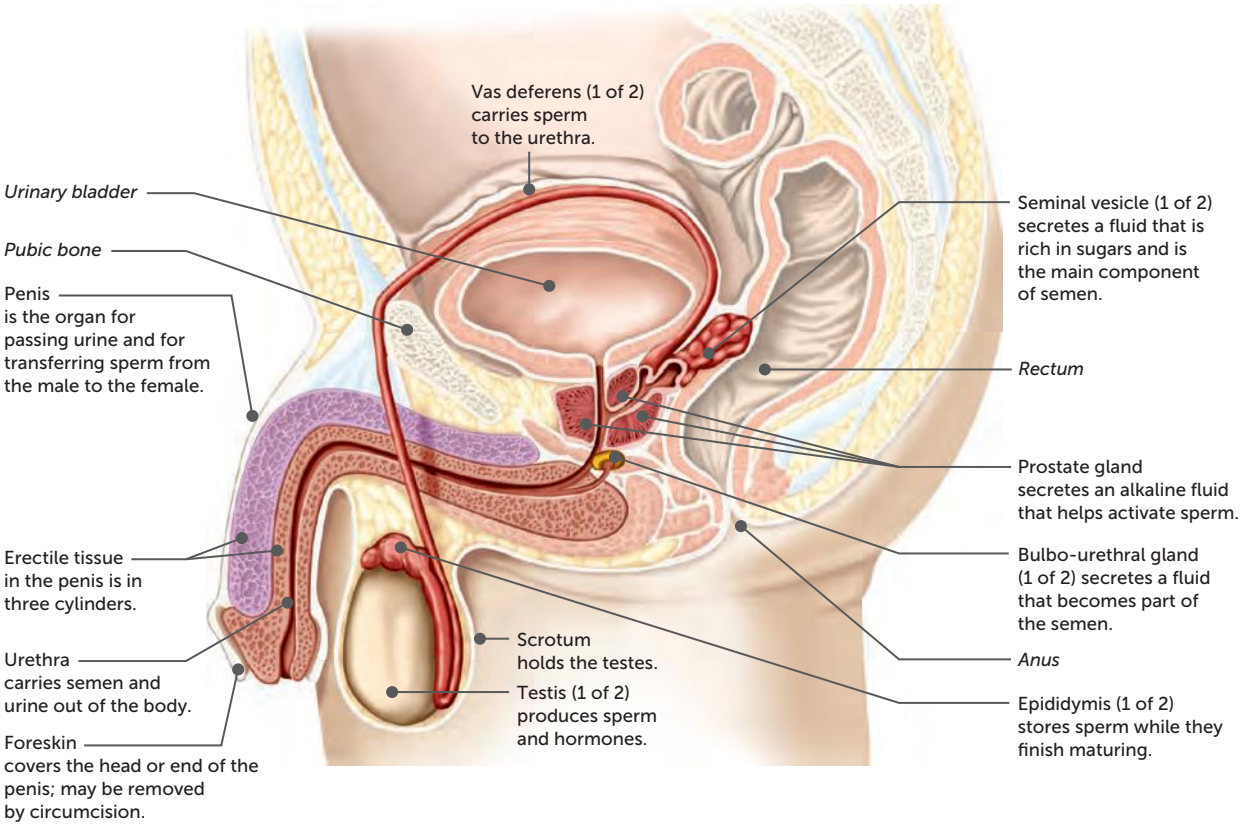


FIGURE 11.4 The male reproductive system. Structures indicated in *italics* are not part of the reproductive system

- The **bulbo-urethral glands**, also known as **Cowper's glands**, are two small yellow glands each about the size of a pea. They are located beneath the prostate on either side of the urethra. They secrete clear mucus, which is carried to the urethra by a duct from each gland. This secretion acts as a lubricant, and much of it precedes the emission of the seminal fluid, with only a small amount included in the semen.

The urethra carries sperm and semen through the penis to a slit-like opening at the tip. In reproduction, the penis is used to transfer sperm from the male to the vagina of the female.

The penis contains connective tissue that has a very rich blood supply. This **erectile tissue** has a large number of sponge-like spaces, which fill with blood during sexual arousal. This causes the penis to enlarge, stiffen and become erect. It is only when the penis is erect that it can be successfully introduced into the vagina.

11.1 Male reproductive system

Male reproductive system
This website gives a brief summary of the male reproductive system.

Key concept

The structure of the male reproductive system allows the production of sperm, and their delivery to facilitate fertilisation of an egg.

Female reproductive system

The primary sex organs, or gonads, of the female are the two ovaries. The **ovaries** produce the female gametes, the **ova** (or **eggs**). Each ovary is an almond-shaped gland approximately 3 cm in length. Unlike the testes, the ovaries are located completely within the body, one on each side of the abdominal cavity supported by ligaments.

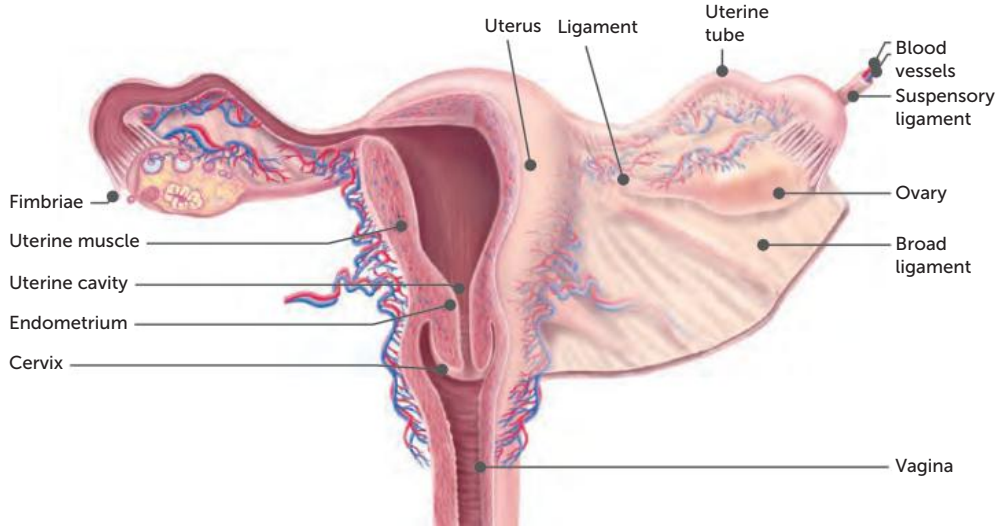
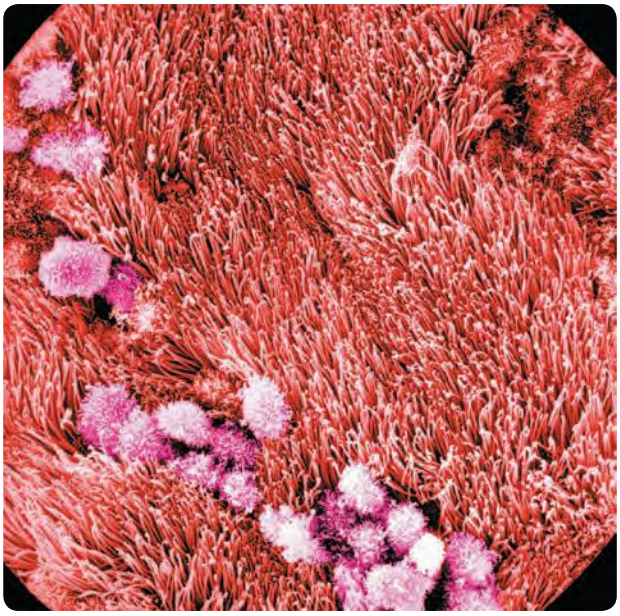


FIGURE 11.5 Front view of the female reproductive system; the left side is cut away to show the internal structures

Each ovary is composed of a mass of connective tissue called the **stroma**. This is surrounded by a layer of cells containing numerous **germ cells**. Each germ cell is enclosed in a **follicle**; at any one time there are numerous follicles in various stages of development. As a follicle matures, it moves to the surface of the ovary and ruptures. The egg is expelled into the funnel-like opening of the **uterine tube**. There are two uterine tubes (also called **Fallopian tubes**, or **oviducts**), one extending from each ovary. They carry the egg from an ovary to the uterus. The funnel-like opening near the ovary is fringed with finger-like projections that appear to just touch the surface of the ovary. These projections, called **fimbriae**, help to guide the egg into the uterine tube. The lining of the tube contains cilia whose movement carries the egg towards the uterus. Contraction of smooth muscles in the wall of the uterine tube also aids this movement of the egg.

9780170449090



Alamy Stock Photo/Phanie

FIGURE 11.6 Transmission electron micrograph (TEM) showing cilia at the surface of an oviduct

The **uterus** (sometimes called the **womb**) is a single, hollow, pear-shaped organ situated behind the urinary bladder and in front of the rectum. It is held in position in the pelvic cavity by broad ligaments that allow some movement. Normally the uterus is tipped forward over the bladder, but variation may occur depending on how full the urinary bladder or the rectum is. The wall of the uterus is made up mainly of smooth muscle, with a soft mucous membrane lining called the **endometrium**. The uterus has a major role to play in protecting and nourishing the developing foetus during pregnancy. You will learn more about this function in Chapter 12.

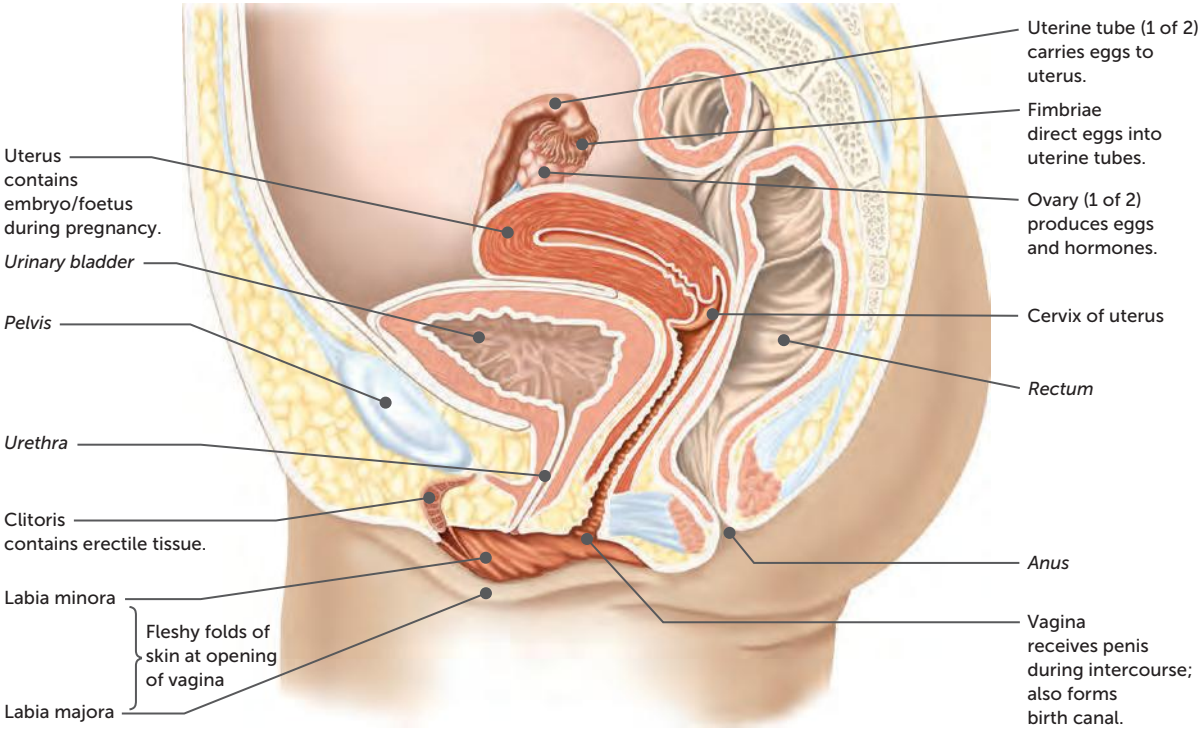


FIGURE 11.7 The female reproductive system as viewed from the side, showing the position of the internal organs. Structures indicated in *italics* are not part of the reproductive system

At the lower end of the uterus is the **cervix**, or neck of the uterus. The cervix protrudes into the **vagina**, a canal leading to the outside of the body that is capable of considerable stretching. The vagina is a muscular structure around 10 cm in length and is lined with mucous membranes. It receives the penis of the male during sexual intercourse and enlarges to form the birth canal during childbirth.

The external opening of the vagina is partially covered by a fold of tissue called the **hymen**. This is stretched and usually torn when sexual intercourse occurs for the first time, but it may be torn by other means. The hymen may also remain in place after sexual intercourse, so its presence or absence is not a reliable sign of virginity.

The vagina opens to the exterior in a region termed the **vulva**, which is made up of the external genital organs of the female – the labia majora, the labia minora and the clitoris.

- The **labia majora** are two fleshy folds of skin, made up of fat and fibrous tissue, containing a large number of glands that produce an oily secretion. Their outer surfaces are pigmented and, after puberty, covered in hair. The inner surfaces are smooth, lack hair, and are moist from the oily secretions.
- Beneath and between the labia majora are two smaller folds of skin, pinkish in colour, without fat, and lacking in pubic hair, the **labia minora**. They surround the space into which the urethra and vagina open.
- At their upper end the labia minora surround the clitoris, a structure equivalent to the penis of the male. The **clitoris** contains erectile tissue, blood vessels and nerves. It is very sensitive to touch, becoming engorged with blood when stimulated.

Female reproductive system
This website gives a brief summary of the female reproductive system.

11.2 Female reproductive system

Activity 11.1
Investigating the female and male reproductive systems

Key concept

The structure of the female reproductive system allows the production of ova, their fertilisation and the development of the resulting offspring.

Questions 11.1

RECALL KNOWLEDGE

1. List the primary sex organs.
2. State the function of each of the following structures of the male reproductive system:
   a. seminiferous tubules
   b. epididymis
   c. interstitial cells of the testis
   d. vas deferens
   e. urethra.
3. List the glands that produce secretions in the semen.
4. State the alternative name for the
   a. egg
   b. uterine tube.
5. List the structures, in order, that the ova pass through from the ovary to the exterior of the body.

APPLY KNOWLEDGE

6. Explain why the scrotum is divided internally.
7. Explain why the muscles in the wall of the scrotum relax during hot weather.
8. Explain the processes that guide the egg through the uterine tubes.

11.2 PRODUCTION OF GAMETES

In Chapter 10 you learnt about the process of meiosis, the type of cell division that produces gametes. Following meiosis, the new cells must develop into the gametes, the sperm or ova. Gamete development, from meiosis to mature gametes, is called **gametogenesis**. There are two types of gametogenesis: formation of spermatozoa in the testis is called **spermatogenesis** and formation of ova in the ovary is called **oogenesis**.

9780170449090

Production of sperm

Spermatogenesis occurs inside the seminiferous tubules of each testis. The seminiferous tubules are lined with immature cells called **spermatogonia**, or sperm mother cells, which contain the diploid number of chromosomes, 46. At puberty, the spermatogonia begin dividing by mitosis and in so doing provide a continuous source of new cells for the production of spermatozoa. Some of the daughter cells from the spermatogonia are pushed inward towards the centre of the tubule, where they undergo a period of growth. These enlarged cells are called **primary spermatocytes**. Primary spermatocytes, like spermatogonia, are diploid and undergo the first stage of meiosis to produce **secondary spermatocytes**. These cells are haploid, as they contain 23 chromosomes. The second meiotic division divides each secondary spermatocyte into two **spermatids**. Thus, four haploid spermatids are formed by meiosis from one diploid spermatogonium.

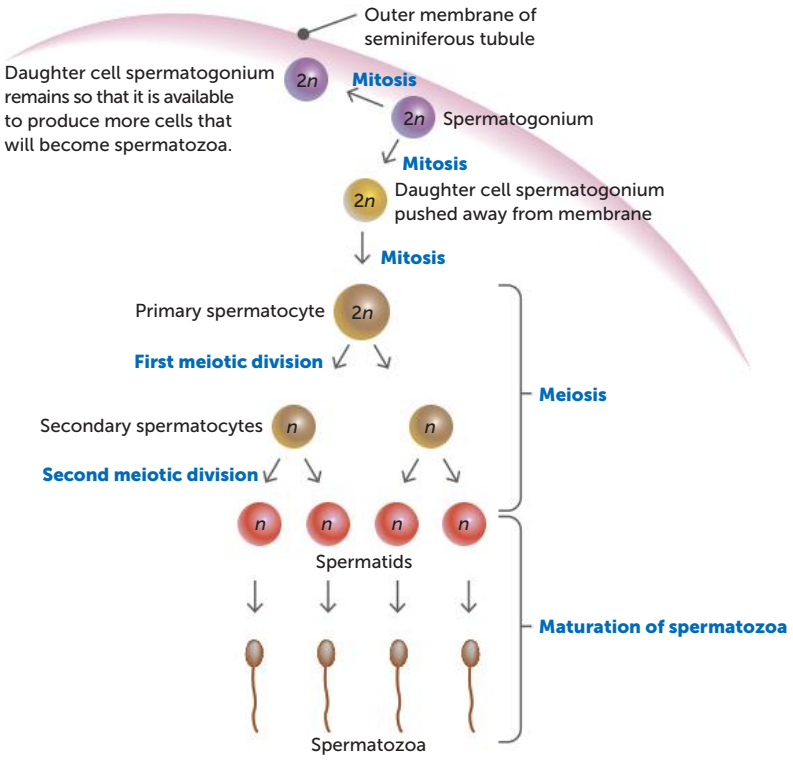


FIGURE 11.8
Spermatogenesis

Spermatogenesis
Click on 'spermatogenesis' at this website to see an animation of the process.

The final stage of spermatogenesis occurs when the spermatids mature into spermatozoa (sperm). During this time, much of the cytoplasm of the cell is lost and a tail containing contractile material forms. The maturing spermatozoa are nourished during this stage by special cells that extend from the outer portion of the seminiferous tubule into the centre. The entire process of spermatogenesis, from spermatogonium to spermatozoa, takes about 72 days and occurs continuously after puberty.

Human sperm are microscopic, being only about 0.06 mm long. Each sperm is made up of a head, neck, middle and tail, as follows:

- The head consists almost entirely of the nuclear material, with a fluid-filled vesicle at its tip, called the acrosome. In the fluid are enzymes, which are important if the sperm reaches an egg. The enzymes break down the layer of cells surrounding the egg so that fertilisation can occur.
- The neck lies between the head and the middle piece.

- The middle piece contains mitochondria, where respiration takes place to provide the sperm with energy for movement. Around the mitochondria is a thin layer of cytoplasm. Because there is so little cytoplasm, sperm have a short survival period and receive their nourishment from the semen in which they are suspended.
- The tail is capable of contractile motions to propel the cell forward.

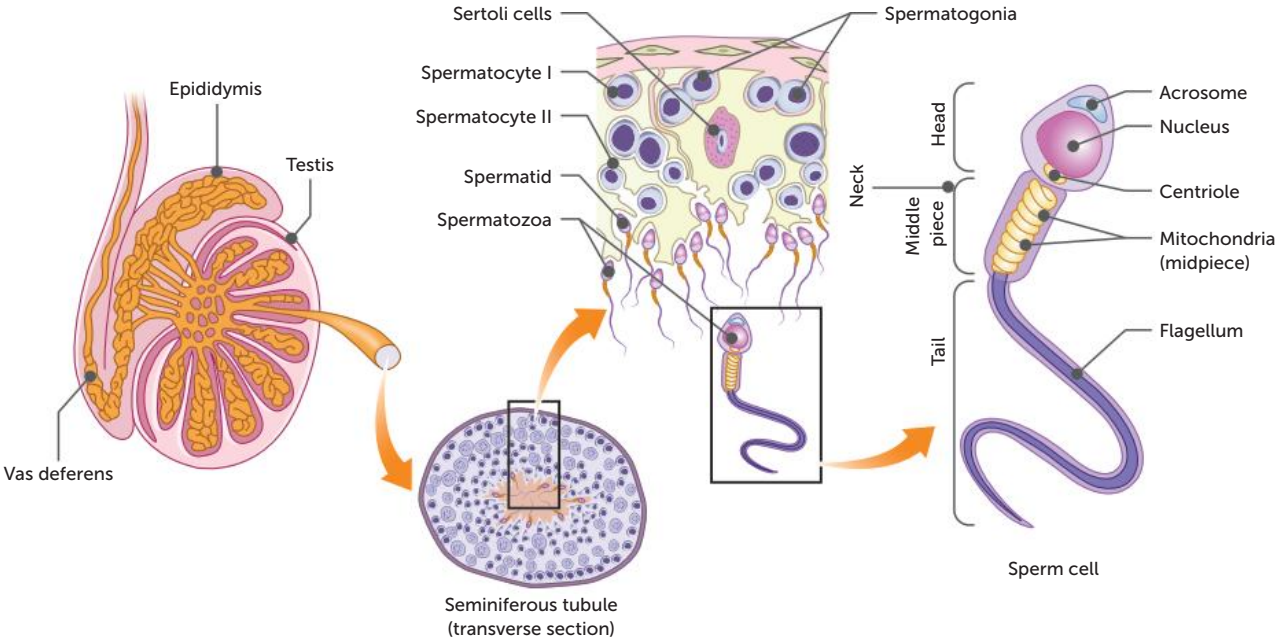
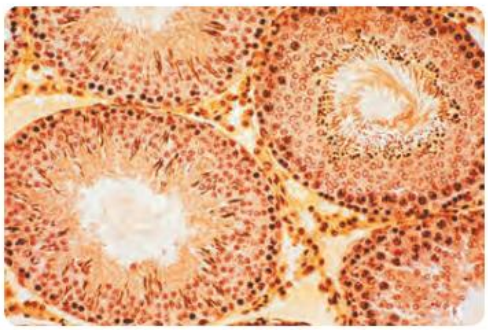
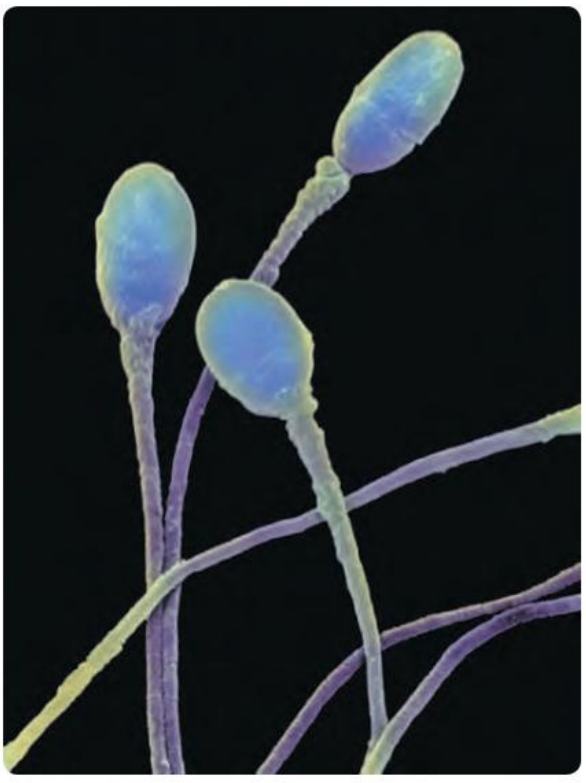


FIGURE 11.9 Section of testis, showing spermatogenesis within a seminiferous tubule



Science Source/Biophoto Associates

FIGURE 11.10 Scanning electron micrograph of a seminiferous tubule



Science Photo Library/Steve Gschmeissner

FIGURE 11.11 Scanning electron micrograph of human spermatozoa

9780170449090

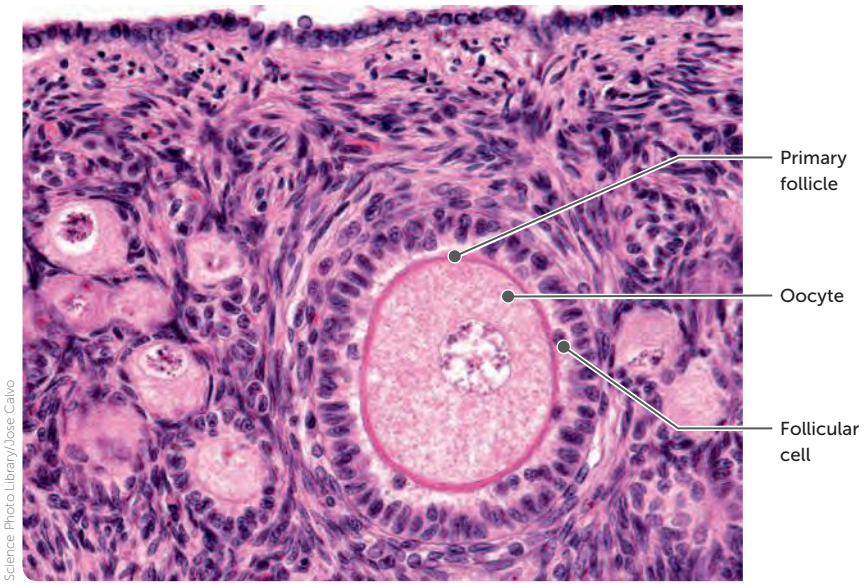
Key concept

Spermatogenesis occurs in the seminiferous tubules of the testes, and results in four spermatozoa from each spermatogonium.

Production of ova

The production of ova within the ovaries is called oogenesis. Like spermatogenesis, oogenesis also involves both meiosis and maturation.

Before a female baby is born, millions of egg mother cells, or **oogonia**, develop in the ovaries. These cells are diploid and divide by mitosis to produce the cells that will eventually develop into ova. By the time of birth, each ovary contains several hundred thousand oogonia, which have undergone a growth phase to become **primary oocytes**. The primary oocytes begin prophase of the first meiotic division, but the process stops at this point, so at birth they are still in the first prophase. Each of these primary oocytes is surrounded by a single layer of cells, forming a **primary follicle**.



Science Photo Library/Jose Calvo

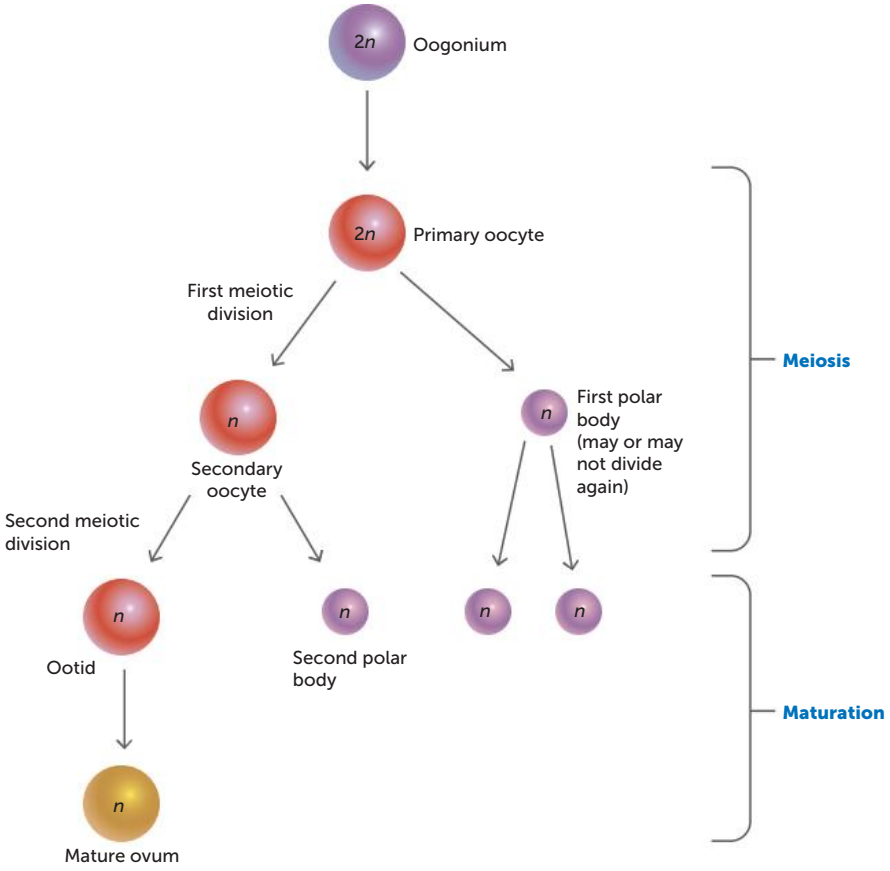
FIGURE 11.12
Stained micrograph of a primary follicle

At puberty, the process of follicle growth and maturation begins. As a follicle matures, the primary oocyte contained within it completes the stages of the first division of meiosis, producing two haploid cells. These cells are of unequal size. The larger one, the **secondary oocyte,** receives half the chromosomes but nearly all the cytoplasm. The smaller cell, the **first polar body**, receives the other half of the chromosomes but very little cytoplasm.

The secondary oocyte immediately commences the second division of meiosis but stops at metaphase. At this stage ovulation occurs: the follicle ruptures, expelling the secondary oocyte along with its **polar body**. The secondary oocyte enters the uterine tube and, if penetrated by a spermatozoon, meiosis is quickly completed. The second division of meiosis also produces two haploid cells of unequal size. The larger one develops into an ovum, or mature egg. The smaller cell is the **second polar body**. The first polar body may also undergo a second meiotic division to produce two additional polar bodies. All polar bodies disintegrate. Thus, in the female, oogenesis produces a single ovum from each primary oocyte; while in the male, spermatogenesis produces four sperm from each primary spermatocyte.

9780170449090

FIGURE 11.13
Oogenesis



Key concept

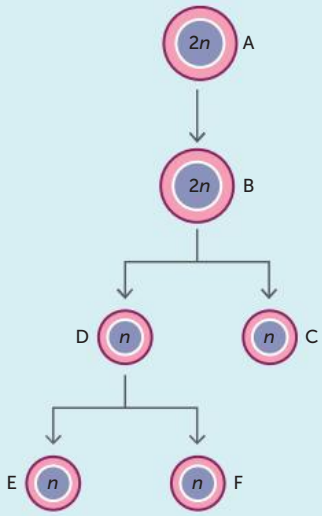
Oogenesis occurs in the ovaries. Oogonia form primary oocytes prior to birth. After puberty, each primary oocyte completes its development to form a secondary oocyte and up to three polar bodies. The secondary oocyte is released during ovulation and completes meiosis if it is fertilised.

Questions 11.2

RECALL KNOWLEDGE

1. List the two types of gametogenesis, and state where they occur.
2. Define each of the following:
   a. spermatogonia
   b. diploid
   c. spermatids
   d. primary follicle
   e. first polar body.
3. Draw a labelled diagram of a sperm.

4 Label the structures on the diagram modelling oogenesis.



5 State how many:
- a sperm are produced from a single spermatocyte
- b ova are produced from a single primary oocyte.

6 Name the structures formed, in order, during spermatogenesis.

APPLY KNOWLEDGE

7 Describe two similarities and two differences between spermatogenesis and oogenesis.

8 Explain the relationship, and differences, between a spermatid and a sperm.

11.3 HORMONAL CONTROL OF THE REPRODUCTIVE SYSTEM

The menstrual and ovarian cycles, and other features of human reproductive systems, depend on the **endocrine glands** for their regulation and control. These are glands that release their secretions, called **hormones**, into the extracellular fluid that surrounds the cells making up the gland. The secretion then usually passes into the capillaries, to be transported by the blood where it circulates until it reaches the target organ – the organ on which it will have an effect. The main target organs for hormones involved in reproductive processes are the testes of the male and the ovaries of the female.

Reproductive hormones

One of the important endocrine glands associated with the reproductive system is the **pituitary gland**. The pituitary is a small organ lying in a pit in the bone below the brain and above the roof of the mouth. Some of the hormones it secretes affect parts of the reproductive system.

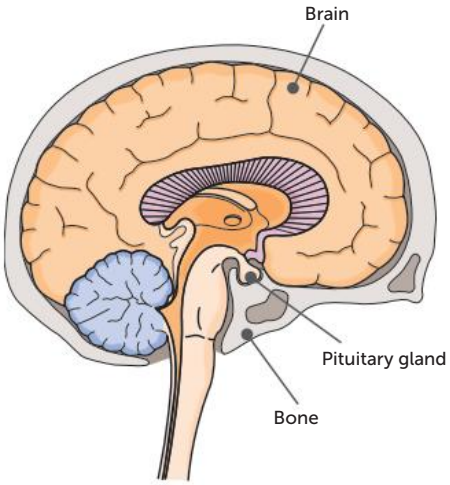


FIGURE 11.14
Location of the pituitary gland

Two hormones secreted by the pituitary affect the gonads and are thus called **gonadotropic hormones**, or **gonadotropins**.

- **Follicle-stimulating hormone (FSH)** stimulates the development and maturation of the ovarian follicle in females. During its development, the ovarian follicle secretes its own hormone, **oestrogen**. Secretion of FSH is reduced as the level of oestrogen increases in the blood.
- **Luteinising hormone (LH)** promotes the final maturation of the ovarian follicle, ovulation, and the formation of the corpus luteum. The corpus luteum secretes another ovarian hormone, **progesterone**, as well as oestrogens. There is a gradual reduction in the production of LH as the level of progesterone in the blood increases.

In addition to the gonadotropic hormones, the pituitary gland releases prolactin and oxytocin. **Prolactin** has a direct effect on the breasts of the woman, and together with other hormones, is important in the preparation and maintenance of milk production. **Oxytocin** causes uterine contractions, promotes the movement of milk in the breast, and has a role in the movement of sperm and the production of testosterone in the testes.

In males, the same gonadotropic hormones are secreted by the pituitary gland. Follicle-stimulating hormone stimulates the epithelial tissue of the seminiferous tubules in the testes to produce sperm. LH stimulates cells in the testes to secrete the hormone testosterone. **Testosterone** is important for the development of immature sperm cells into mature spermatozoa, and has a major role in the maintenance of the male reproductive organs and sex drive.

At puberty, the secretion of gonadotropic hormones stimulates a number of changes, both physical and psychological and in both males and females. In males, the production of testosterone influences the development of the body to sexual maturity. In females, sexual maturation is brought about by oestrogens.

WS

11.3 Female hormones

Your hormones
This website has more information about the different hormones.

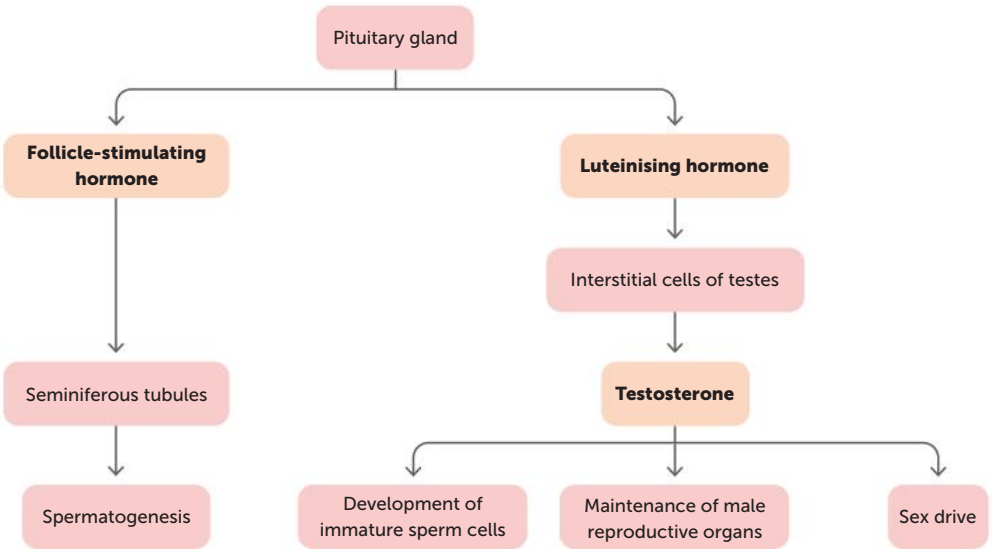


FIGURE 11.15
Summary of how male reproductive hormones influence the production of sperm

The ovarian cycle

The **ovarian cycle** is a series of events that take place within the ovaries. It includes the maturation of an egg and its release into a uterine tube. Associated with these events are the development of follicles in the ovary and the formation of a structure called the corpus luteum. The length of the ovarian cycle is highly variable, depending on the individual and her circumstances. It may range from 20 to 40 days, with an average of about 28 days. For this reason, the ovarian cycle is commonly considered to be a 28-day cycle, even though only about 30% of women have a cycle that is 27 or 28 days in length.

When a female matures sexually at **puberty**, some of the primary follicles which contain a primary oocyte undergo further development. During the first half of the ovarian cycle the levels of follicle-stimulating hormone and luteinising hormone slowly increase, prompting the growth and maturation of follicles over a 10–14 day period. Each follicle that develops goes through the following steps.

1. Cells forming the wall of the primary follicle begin to enlarge and divide, creating a layer of cells around the developing oocyte.
2. Secretions from these cells create a fluid-filled space that gradually forces the oocyte to the edge of the follicle. It is now referred to as a **secondary follicle**.
3. As more fluid accumulates within the follicle, it continues to enlarge and gradually moves towards the surface of the ovary.
4. On reaching the surface, it produces a bulge that looks like a swollen blister on the surface of the ovary. At this stage it is referred to as a **mature follicle** (previously known as a **Graafian follicle**).

Several secondary follicles may commence development in each ovarian cycle, but usually only one completes development. The others normally break down to be reabsorbed into the ovary.

As the follicles grow, they release oestrogen, and a low level of progesterone. At approximately day 14, the level of oestrogen is high enough to stimulate a spike in LH and FSH. The surge in LH causes the ovulation of the most mature follicle, while the other follicles degenerate, releasing less oestrogen.

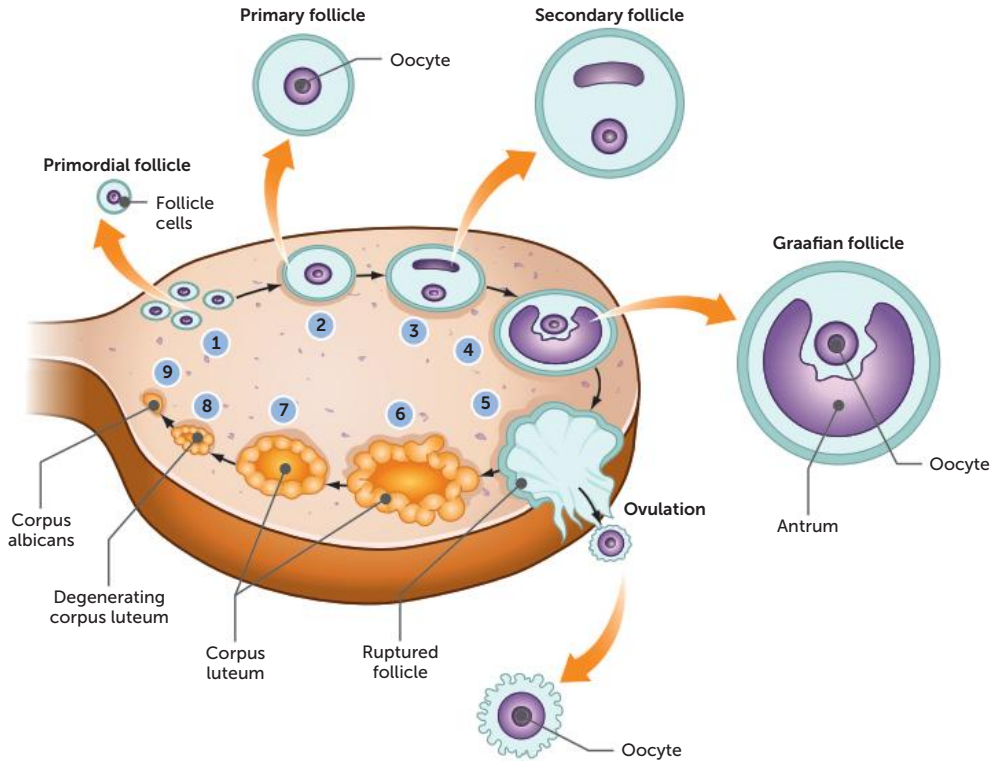


FIGURE 11.16 The ovarian cycle

At ovulation, the mature follicle bursts and expels the oocyte. The open end of the uterine tube is like a funnel over the ovary. Beating cilia within the funnel create a current that sweeps the egg into the uterine tube. Usually only one follicle matures at a time, so only one oocyte is released. However, very occasionally, two or more follicles may burst at the same time, releasing more than one oocyte. The oocyte is gradually swept down the uterine tube towards the uterus by the beating of cilia lining the inside of the tube.

Following ovulation, the ruptured follicle collapses and the blood within forms a clot. The clot is gradually absorbed by the remaining follicle cells, which enlarge and change colour to form a cream-coloured body, the **corpus luteum** (Latin for 'yellow body'). The corpus luteum secretes oestrogen and progesterone. Progesterone influences the development of the lining of the uterus, preparing it for implantation if fertilisation occurs. It also inhibits the release of FSH and LH which prevent other follicles developing. If fertilisation has not occurred, the corpus luteum reaches its maximum development about 8 to 10 days after ovulation. It then begins to degenerate into a fibrous mass of scar tissue, the **corpus albicans** (Latin for 'white body'), which eventually disappears. Another ovarian cycle then begins due to the reduction in progesterone and oestrogen. It seems to be a matter of chance whether it occurs in the same ovary or in the opposite ovary.

If fertilisation of an egg takes place and pregnancy follows, the corpus luteum continues to develop and the ovarian cycles cease. The corpus luteum is maintained by **human chorionic gonadotropin (HCG)**, a hormone produced by the developing placenta in a pregnant woman. Once the placenta is itself able to secrete oestrogens and progesterone, the corpus luteum begins to degenerate. (The role of the placenta is discussed in more detail in Chapter 12.) Degeneration is slow, and the corpus luteum is still present in the ovary at childbirth. Ovarian cycles usually resume only after breastfeeding of the baby has ceased.

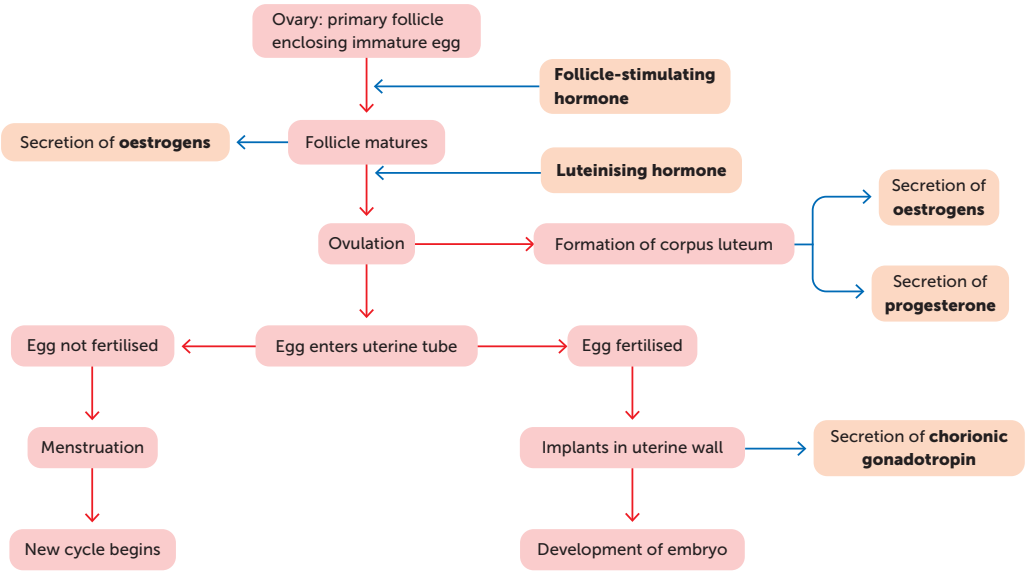


FIGURE 11.17
Summary of the relationship between the ovarian cycle and the reproductive hormones

Activity 11.2
Observing the ovary

The menstrual cycle

While the ovarian cycle is occurring, the uterus and, to a lesser extent, the vagina also go through a series of changes called the **menstrual cycle**. The changes in the uterine lining, the endometrium, are in preparation for a developing embryo in case the egg released at ovulation is fertilised.

In the first stage of the menstrual cycle, while the follicle is maturing in the ovarian cycle, progesterone causes the endometrium of the uterus to become thicker and softer. There is also an increase in the number of blood vessels and mucus-secreting glands. After ovulation the endometrium continues to thicken, and glands within it begin to secrete a watery mucus rich in glycogen.

9780170449090

If the egg is not fertilised by a sperm, the corpus luteum degenerates, reducing the amount of progesterone, which results in its breakdown. About 14 days after ovulation, blood from broken-down capillaries, mucous secretions and cell debris from the uterine lining are lost through the vagina. This is **menstruation**. Menstruation takes place over several days and is commonly referred to as the **menstrual period** (or, often, just 'period'). As this event is the most recognisable point in the menstrual cycle, the onset of menstruation is taken to be day 1 of the cycle. A summary of the major stages of the menstrual cycle is given in Table 11.1.

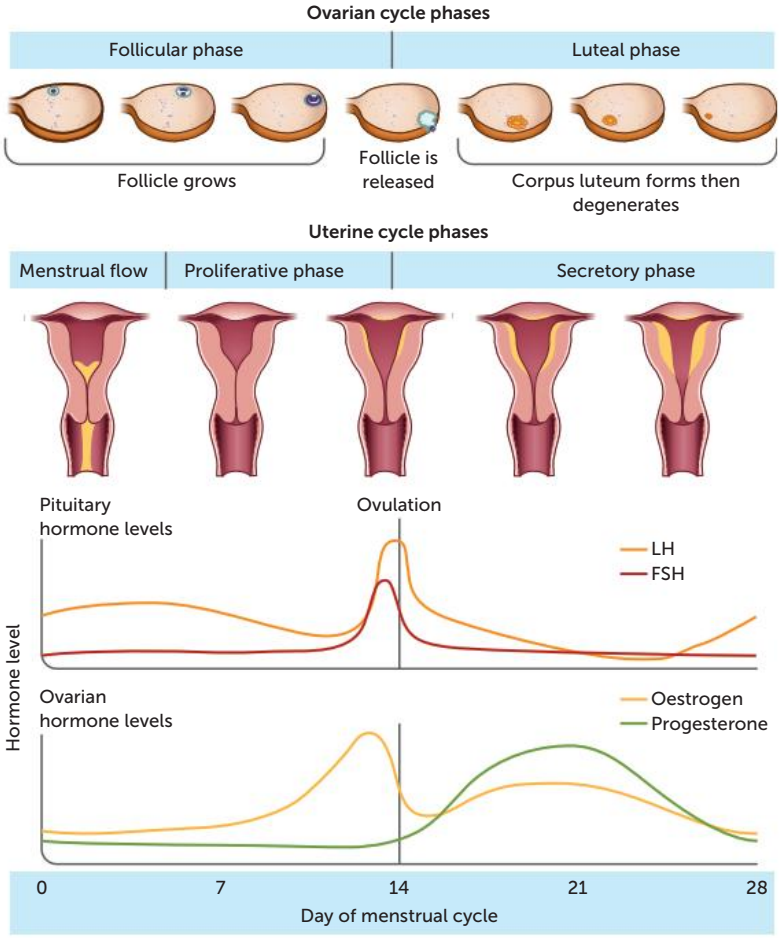


FIGURE 11.18 Correlation between the menstrual and ovarian cycles

TABLE 11.1 Major stages of the menstrual cycle

STAGE	AVERAGE TIME SPAN (DAYS)	EVENTS
Menstruation	1–4	Uterine bleeding, accompanied by shedding of the endometrium
Preovulation	5–12	Endometrial repair begins; development of ovarian follicle; uterine lining gradually thickens
Ovulation	13–15	Rupture of mature follicle, releasing egg
Secretion	16–20	Secretion of watery mucus by glands of endometrium, cervix and uterine tubes; movement and breakdown of unfertilised egg; development of corpus luteum
Premenstruation	21–28	Degeneration of corpus luteum; deterioration of endometrium

A female's first menstruation is called **menarche** and marks the commencement of puberty. From that time on, she will have a menstrual cycle about once a month unless it is interrupted by pregnancy. The cycles last until **menopause**, the time when the processes that occurred at puberty are reversed. These changes usually begin between the ages of 45 and 55 and take place over a

period of years, during which time the menstrual cycle becomes irregular until it eventually ceases. Typically, a woman has a potential of about 35 child-bearing years, and in most women only about 400 of the initial 400 000 potential eggs reach maturity.

Puberty and the development of secondary sexual characteristics

The secretion of sex hormones at puberty brings about the development of **secondary sexual characteristics** – those characteristics associated with a person's sex but not directly involved in sexual reproduction. In females, the development of secondary sexual characteristics begins with enlarging of the breasts. At the same time, there is a broadening of the hips. The growth of the pelvic bones and the deposition of fat contribute both to this and to the more rounded contours of the female body compared with those of the male.

The distribution of added hair and change in voice properties are also secondary sexual characteristics. Pubic hair begins to grow early in puberty in both males and females. As puberty progresses, it changes from straight and fairly light in colour to a thicker, curlier and darker covering. Shortly afterwards, hair grows in the armpits of both sexes. Later still in males, it grows on the face, chest and possibly the back. Males also have an increase in the size of the larynx, with an associated lengthening of the vocal cords. This results in a gradual deepening of the voice, even though it may appear to 'break' suddenly. In females the voice deepens to some extent but seldom reaches the deeper tones of the male.

Key concept

The periodic changes of the ovaries (ovarian cycle) and the endometrium (menstrual cycle) are regulated by hormones.

Summary of reproductive hormones

Table 11.2 summarises the hormones involved in reproduction.

TABLE 11.2 Reproductive hormones

HORMONE	TARGET ORGAN	EFFECT OF HORMONE
Follicle-stimulating hormone (FSH) from pituitary gland	Seminiferous tubules of testes	Production of sperm
	Follicles of ovaries	Maturation of ovarian follicles
Human chorionic gonadotropin (HCG) from placenta	Corpus luteum	Maintenance of corpus luteum during early stages of pregnancy
Lactogenic hormone (prolactin) from pituitary gland	Breasts	Production of milk in activated glands
Luteinising hormone (LH) from pituitary gland	Interstitial cells of testes	Stimulates secretion of testosterone
	Cells of the ovaries	Stimulates secretion of oestrogens and progesterone
Oestrogens from ovarian follicle and corpus luteum	Various	Development of female reproductive system
		Development of secondary sexual characteristics
Oxytocin from pituitary gland	Uterus	Stimulates contraction of smooth muscle
	Breasts	Promotes contraction of muscle cells surrounding breast lobules
Progesterone from corpus luteum	Uterus	Maintenance of endometrium
	Placenta	Development and maintenance of placenta
	Breasts	Development of milk-secreting glands
Testosterone from cells in testis	Various	Development of male reproductive system
		Development of secondary sexual characteristics

9780170449090

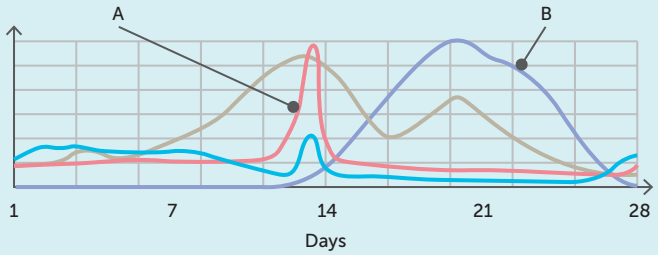
Questions 11.3

RECALL KNOWLEDGE

1 How are hormones transported around the body?
2 Name the gonadotrophic hormones.
3 State the effect of:
   a follicle-stimulating hormone
   b luteinising hormone
   c prolactin
   d oxytocin
   e progesterone
   f oestrogen
   g testosterone.
4 How long is the ovarian cycle?
5 Describe the events of the ovarian cycle.
6 Describe a corpus luteum.
7 With reference to the ovarian cycle, when does menstruation usually start?
8 List the secondary sexual characteristics of:
   a males
   b females.

APPLY KNOWLEDGE

9 Name the hormones labelled A and B on the graph below. Explain how you determined the answer.



10 Explain the difference between a primary follicle and a secondary follicle.
11 The ovarian cycle ceases during pregnancy. Explain how this is regulated.
12 Suggest why menstruation needs to happen in female humans.
13 Explain how the body ensures that ovulation takes place when a follicle is large enough.

9780170449090

CHAPTER 11 ACTIVITIES

ACTIVITY 11.1 Investigating the female and male reproductive systems

The reproductive systems of most mammals are similar in structure and function. Therefore, examination of the reproductive structures of rats will help you to understand the human reproductive system.

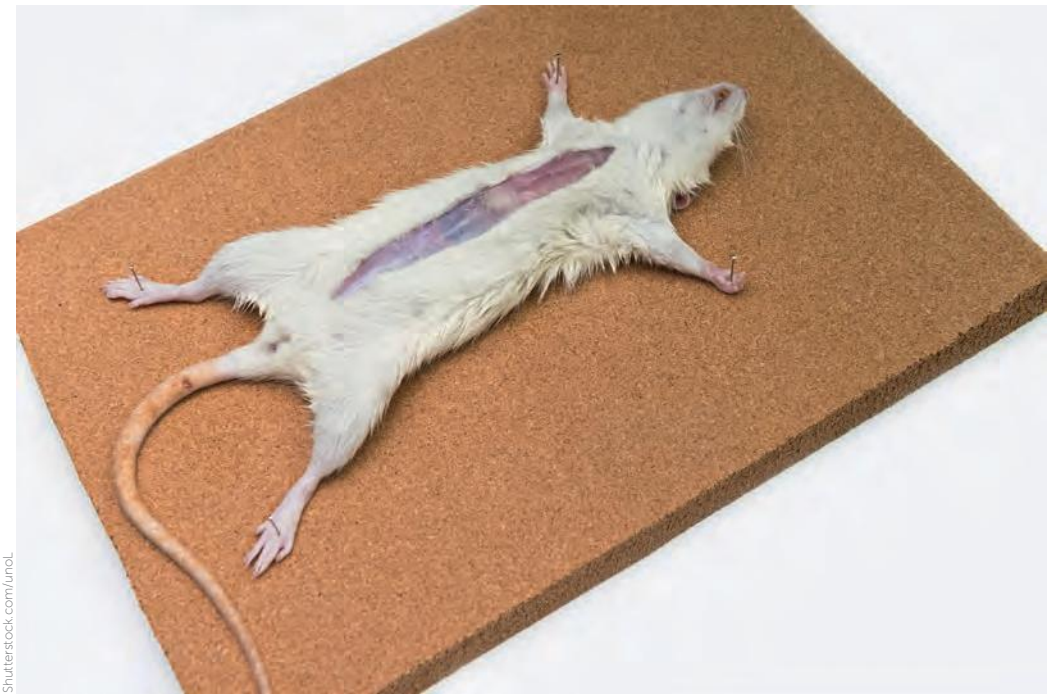
Your teacher may ask you to dissect a rat yourself, may demonstrate the dissection, or may refer you to a video or photographs for this activity.

You will need (if doing the dissection yourself)

A female and a male rat; dissecting board; dissecting instruments; string; hand lens or magnifying glass; disposable gloves

What to do

1 Tie the rat to the dissecting board so that its legs are lying away from its body, as shown below.



Shutterstock.com/unoL

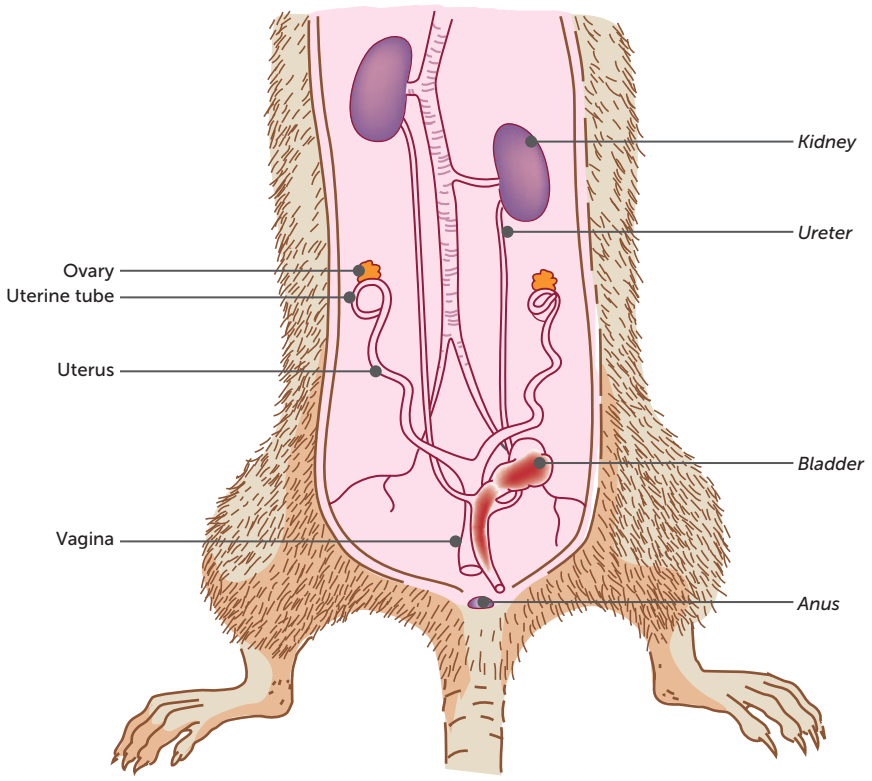
Female rat

2 Identify the external features of the rat that are associated with reproduction. In the female rat, locate the genital opening and the mammary glands. In addition, locate the urethra and the anus.

3 Count the number of nipples on the underside of the abdomen.

4 Follow your teacher's instructions to open the body cavity to reveal the reproductive organs. There may be some fat associated with these organs, but do not try to remove it. You may just need to displace it so the reproductive organs can be easily observed.

5 Locate the vagina.
6 Locate the two uteri that extend from the vagina up each side of the body cavity.
7 At the anterior (front) end of each uterus is a very short uterine tube that you may find difficult to identify.
8 At the end of each uterus is a small, round, orange-coloured structure. This is the ovary.
9 It may help to insert a blunt seeker into the vagina to trace the pathway that sperm would take.
10 Identify the urinary bladder. If the rat is not preserved, this will appear as a semi-transparent bag containing clear fluid.



Reproductive system of a female rat, with alimentary canal removed

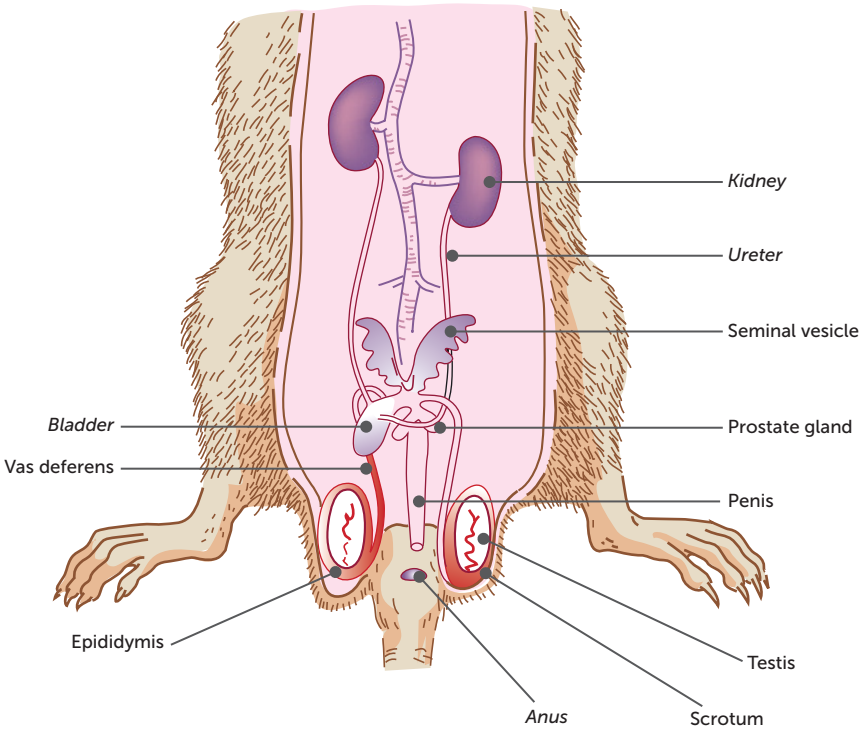
Male rat

11 Identify the external features of the rat that are associated with reproduction. In the male rat, locate the scrotum containing the testes, the penis and the opening of the urethra at the tip of the penis.
12 Follow your teacher's instructions to open one of the scrotal sacs to reveal the testis. This will involve cutting through the skin around the base of the penis and continuing the cut down through the middle of one of the scrotal sacs. Use a blunt probe to gently lift the testis clear of the scrotal sac.
13 You will now be able to identify the vas deferens and the epididymis, as well as the testis. Use a hand lens, or magnifying glass, to see the seminiferous tubules inside the testis and the tubules that make up the epididymis.
14 Clear the skin away from the rear portion of the belly of the rat. Tracing the vas deferens forward, cut through the wall of the rat's abdomen so that you can see where it enters the body cavity.

9780170449090

15 Identify the urinary bladder. If the rat is not preserved, this will appear as a semi-transparent bag containing clear fluid. Near the bladder, the two vasa deferentia, one from each testis, join together to form the urethra.

16 Near the point where the vasa deferentia join, you will be able to see two white, elongated glands with a crinkly appearance. These are the seminal vesicles. The prostate gland, although present in the rat, is very difficult to see.



Reproductive system of a male rat, with alimentary canal removed

Studying your observations

1 Draw a diagram of your dissections, labelling all the structures that you have identified.

2 On your diagram, use arrows to show:
   - **a** the path an egg would take after it has been expelled from the ovary.
   - **b** the path that the sperm and semen would follow as they travel to the tip of the penis
   - **c** the path that you think the sperm and semen would take after they have been deposited in the vagina.

3 List any differences between the reproductive structures of a female rat and a female human.

4 Of the differences you have noted, which ones are related to the rat having a number of offspring at one time?

5 List any differences between the reproductive structures of a male rat and a male human.

9780170449090

ACTIVITY 11.2 Observing the ovary

This activity allows you to examine the various stages of the ovarian cycle. Your teacher may have set up a demonstration slide for you to view.

You will need (if doing this activity yourself)

A microscope; prepared microscope slides of a transverse section of a mammalian ovary

What to do

1. Set up the microscope and place a prepared slide on the stage. Examine the cross-section of an ovary.
2. Identify the various stages of development of the follicles using Figure 11.16 on page 287.

Studying your observations

1. Draw a diagram of what you viewed on the slide, labelling all visible structures.
2. Compare your drawing with Figure 11.16. If there were there any structures you were unable to see on the microscope slide, annotate your diagram using information from Figure 11.16.

9780170449090

CHAPTER 11 SUMMARY

- Humans reproduce by a sperm and an egg fusing to form a zygote.
- Sperm are produced in the testes, which are located in the scrotum.
- The testes are divided into lobules, which are filled with the seminiferous tubules. These tubules join to form the epididymis, which leave each testis and become the vas deferens.
- Testosterone is produced by the interstitial cells found between the seminiferous tubules.
- Semen contains fluids from the seminal vesicles, prostate gland and bulbo-urethral gland. It nourishes the sperm and aids their transport.
- The sperm exits the male via the urethra, entering the female vagina during sexual intercourse.
- Ovaries contain germ cells in a follicle. When the follicle matures it ruptures, releasing the egg into the uterine tube so that it can travel to the uterus.
- The uterus is separated from the exterior by the cervix, which opens to the vagina and then the vulva.
- Gametogenesis is the production of gametes by meiosis.
- Sperm are produced by spermatogenesis, which occurs in the seminiferous tubules.
- The seminiferous tubules are lined with spermatogonia. These are diploid cells and divide by mitosis once puberty starts.
- Some of the daughter cells move to the centre of the tubule where they grow and become primary spermatocytes. These start meiosis and form secondary spermatocytes. After the second meiotic division the cells are called spermatids.
- Spermatids form mature spermatozoa (sperm) by forming a mobile tail and losing most of the cytoplasm.
- Sperm are made up of a head, neck, middle piece and tail.
- Eggs, or ova, are produced by oogenesis.
- Before birth, there are millions of diploid oogonia in the ovaries. These undergo growth to form primary oocytes, which begin the prophase of the first meiotic division.
- At puberty, the meiotic division continues for a primary oocyte, forming a secondary oocyte and a polar body. The secondary oocyte enters the second stage of meiosis, but stops at the metaphase until fertilisation.
- The ovarian and menstrual cycles are controlled by the endocrine system.
- Follicle-stimulating hormone and luteinising hormone are gonadotropins that are produced by the pituitary gland.
- Follicle-stimulating hormone stimulates the development and maturation of the follicles in females and epithelial cells to produce sperm in males.
- Luteinising hormone promotes the final maturation of the follicle, ovulation and the formation of the corpus luteum in females and the production of testosterone in males.
- Ovarian follicles secrete oestrogen, while the corpus luteum secretes progesterone and oestrogen.
- Testosterone leads to the maturation of spermatozoa in addition to the maintenance of the reproductive organs and sex drive.
- During the ovarian cycle the outer cells form a layer around the developing egg. They produce a fluid that pushes the egg to the edge, forming a secondary follicle. When the egg reaches the edge it pushes against the surface; the follicle is now called a mature follicle.
- The mature follicle bursts at ovulation, releasing the egg into the uterine tube.
- A corpus luteum forms in the ruptured follicle. If fertilisation does not occur, it degenerates to become a corpus albicans. If fertilisation does occur, the

9780170449090

placenta produces human chorionic gonadotropin, which maintains the corpus luteum until the placenta is able to produce oestrogen and progesterone.

- During the menstrual cycle, progesterone from the developing follicle causes the endometrium to thicken and soften. If the egg is not fertilised, the reduction in progesterone leads to the breakdown of the endometrium. The blood, mucus and cells are lost through the vagina during menstruation.
- Menarche is when a female starts menstruating, and menopause is when she stops menstruating.
- The secondary sexual characteristics are associated with a person's sex, but not with reproduction – for example, breast development in females and a deepening of the voice in males.

9780170449090

CHAPTER 11 GLOSSARY

Bulbo-urethral gland One of a pair of small yellow glands that secrete a lubricating fluid into the urethra; also called Cowper's gland

Cervix The neck of the uterus, leading into the vagina

Clitoris The erectile organ of the female; it is equivalent to the penis in males

Corpus albicans A fibrous mass of scar tissue left on the ovary after the corpus luteum degenerates

Corpus luteum The temporary endocrine gland that forms in the ovary after the release of an egg

Cowper's gland *see* bulbo-urethral gland

Egg *see* ova

Endocrine gland A gland that secretes hormones directly into adjacent tissue; also called a ductless gland

Endometrium The soft mucous membrane lining the uterus

Epididymis A highly folded tubule behind each testis in which the sperm mature

Erectile tissue Spongy tissue in the penis that fills with blood to produce an erection

Fallopian tube *see* uterine tube

Fimbriae The finger-like projections of the uterine tube

First polar body The smaller of two unequal cells formed when the primary oocyte divides during the first division of meiosis

Follicle A small secretory sac or gland

Follicle-stimulating hormone (FSH) A hormone that stimulates the development of a follicle in the ovary

Gametogenesis The formation and development of the gametes

Germ cell Cells in the ovary that are able to develop into ova

Gonadotropic hormones *see* gonadotropins

Gonadotropins Hormones that affect the sex organs; also called gonadotropic hormones

Graafian follicle *see* mature follicle

Hormone A chemical secreted by an endocrine gland, often carried in the blood; it affects the functioning of a cell or organ

Human chorionic gonadotropin (HCG) A hormone produced by the placenta during pregnancy

Hymen The fold of tissue that covers the external opening of the vagina

Interstitial cells Cells located in the mature testis; they secrete testosterone

Labia (labia majora and labia minora) The fleshy folds of skin lining the opening to the vagina and urethra

Lobule A compartment within the testis that contains seminiferous tubules

Luteinising hormone (LH) A hormone that promotes final maturation of the ovarian follicle and formation of the corpus luteum

Mature follicle A fluid-filled structure in the ovary; it contains an immature egg and its surrounding tissues

Menarche The onset of menstruation

Menopause Cessation of menstruation

Menstrual cycle The regular series of changes that take place in the walls of the uterus of a non-pregnant female

Menstrual period *see* menstrual cycle

Menstruation The periodic discharge of blood and tissue fluid due to the breakdown of the lining of the uterus; also known as the menstrual period

Oestrogen A general name for a female sex hormone; it develops or maintains female reproductive structures

Oogenesis The formation and development of the ovum within the ovary

Oogonia The cells in the ovaries that produce primary oocytes by mitosis; singular: oogonium

Ova Mature egg cells; singular: ovum

Ovarian cycle The regular series of events that take place within an ovary of a non-pregnant female, associated with the maturation of an egg

Ovaries The organs in which the female gametes, the ova (or eggs), are produced

Oviduct *see* uterine tube

9780170449090

Oxytocin A hormone produced by the hypothalamus and released by the posterior pituitary gland that has a role in childbirth and lactation

Pituitary gland An endocrine gland located below the brain

Polar body A haploid cell produced during oogenesis that contains a nucleus and very little cytoplasm

Primary follicle A dormant egg within the ovary

Primary oocyte Diploid cells in the ovary that undergo meiosis to produce haploid secondary oocytes

Primary sex organs The organs that produce gametes: testes and ovaries

Primary spermatocyte Diploid cells that undergo the first division of meiosis to produce secondary spermatocytes

Progesterone A female sex hormone produced by the ovaries; it helps prepare the uterine lining for a fertilised egg and the mammary glands for milk secretion

Prolactin A hormone that promotes milk production during and after pregnancy

Prostate gland A gland that surrounds the urethra just below the bladder; it secretes a fluid that becomes part of the semen

Puberty The period during which a person becomes sexually mature

Scrotum The pouch outside the abdominal cavity in which the testes are located

Second polar body A polar body formed during the second stage of meiosis

Secondary follicle A previously dormant (primary) follicle that has begun to develop

Secondary oocyte Haploid cells that undergo meiosis to produce ova

Secondary sex organs Organs essential for reproduction but which do not produce gametes

Secondary sexual characteristics A characteristic associated with an individual's sex but which is not involved in sexual reproduction

Secondary spermatocyte Haploid cells that undergo the second division of meiosis to produce spermatids

Semen The liquid that nourishes and aids the transport of sperm; also called seminal fluid

Seminal fluid *see* semen

Seminal vesicles A pair of pouch-like organs that secrete a thick fluid which is a major part of semen

Seminiferous tubules A tightly coiled duct, located in the testis, where sperm are produced

Sperm *see* spermatozoa

Sperm duct *see* vas deferens

Spermatids Haploid cells produced from spermatogonia by meiosis; they mature to form spermatozoa

Spermatogenesis The formation and development of the spermatozoa

Spermatogonia Immature cells lining the seminiferous tubules

Spermatozoa Male gametes; also called sperm; singular: spermatozoon

Stroma Connective tissue in the ovary

Testes The male sex organs that produce sperm and the hormone testosterone; singular: testis

Testosterone The male sex hormone secreted by endocrine cells within a mature testis

Urethra The duct that carries urine from the bladder to the exterior; in males it also carries semen

Uterine tube The tube that carries the eggs from the ovaries to the uterus; also called a Fallopian tube or oviduct

Uterus The hollow, pear-shaped organ situated between the urinary bladder and the rectum in females; also called the womb

Vagina The canal leading from the uterus to the exterior of the female body

Vas deferens The tube that carries the sperm away from the testis; also called the sperm duct; plural: vasa deferentia

Vulva Female external genitalia

Womb *see* uterus

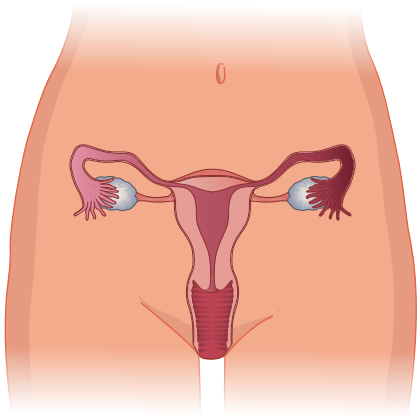
Zygote The fertilised egg from which a new individual develops

9780170449090

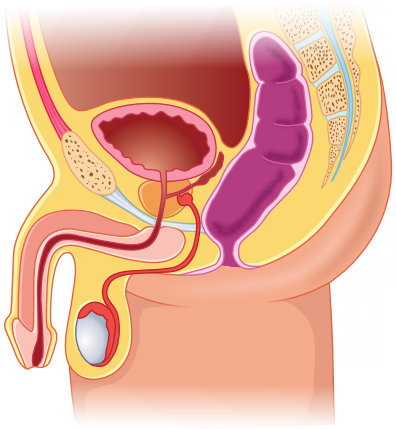
CHAPTER 11 REVIEW QUESTIONS

Recall

1 Label the uterus, cervix, ovary, endometrium, uterine tube and vagina on the diagram below.



2 Label the testis, scrotum, penis, vas deferens, epididymis, urethra, prostate gland, bulbo-urethral glands and seminal vesicles on the diagram below.



3 Describe the internal structure of a testis, including the location of sperm production.

4 Describe how the ovaries are held in position in the pelvic cavity.

5 State the function of each of the following structures:
- a uterine tubes
- b cervix
- c seminiferous tubules
- d prostate gland
- e urethra
- f endometrium.

6 a Define 'target organ'.
- b What are the target organs for testosterone and follicle-stimulating hormone?

7 What do the terms 'menarche' and 'menopause' refer to?

8 a What are secondary sexual characteristics?
- b Briefly describe the development of secondary sexual characteristics in both males and females.

9 List the following events in the order in which they would occur in the female body: ovulation; the endometrium begins to thicken; formation of the corpus luteum; a follicle begins to develop; uterine bleeding; egg travels down the uterine tube; follicle approaches maturity; degeneration of the corpus luteum; breakdown of unfertilised egg; development of the mature follicle; deterioration of the endometrium.

10 What are the primary sex organs of the male? What are their functions?

11 a What are gametes?
- b Draw a sperm and identify the main parts.
- c Outline the events that take place in spermatogenesis and oogenesis.
- d List the differences between the two processes.

12 List the glands that secrete seminal fluid and describe the function of each.

13 a List the stages of the ovarian cycle, using a diagram to illustrate your answer.
- b Describe ovulation.
- c Describe the changes that the corpus luteum undergoes during a normal ovarian cycle.
- d How do these changes differ if pregnancy occurs?

9780170449090

Explain

14 Why is reproduction necessary for the human species?

15 The location of the testes within a scrotum makes them vulnerable to damage. Explain the reason for their location.

16 The seminiferous tubules are highly coiled, and if they were stretched out would be about 800 m in length. Why is such a great length required in the tubules? List as many advantages as you can to support your answer.

17 Describe how follicle-stimulating hormone and luteinising hormone regulate the male reproductive system.

18 Explain the role of hormones in regulating the ovarian and menstrual cycles.

Apply

19 Draw a diagram of the female reproductive system and mark in the:
- **a** place where sperm are deposited
- **b** site where fertilisation takes place
- **c** path taken by the sperm to unite with the egg
- **d** path the egg follows to unite with the sperm.

20 **a** It has been found that men who regularly wear tight-fitting underwear may produce fewer sperm and have reduced sperm quality. Suggest why this may be so.
- **b** If a man uses a laptop computer on his lap for long periods, is it possible that his sperm production could be affected? Explain why this can happen.

21 Some oral contraceptive pills contain oestrogen and progesterone. Suggest how they are able to prevent ovulation.

Extend

22 The head of the penis is covered in loose skin called the foreskin. Removal of the foreskin is an operation called circumcision. Circumcision was once a widespread practice in Australia but is now rarely performed. Find out:
- **a** the advantages of circumcision
- **b** any disadvantages associated with circumcision and why most medical practitioners are now opposed to the practice
- **c** whether circumcision has links with specific cultures or particular religious beliefs.

23 Endometriosis is a condition where the endometrium, the tissue that lines the uterus, grows outside the uterus in other parts of the body. Most growths occur on organs in the abdominal cavity such as the ovaries, uterine tubes, and the outside of the uterus, bladder and intestines. The growths detach and bleed at menstruation, but the blood and tissue cannot be passed to the outside. It is estimated that at least 10% of women suffer from endometriosis during their reproductive years. About 30–40% of those sufferers are infertile; it is one of the main causes of infertility. Find out:
- **a** the symptoms of endometriosis
- **b** possible causes of endometriosis
- **c** how diagnosis is made
- **d** the treatment.

24 In the 1830s, the average age of a female's first menstruation was 17. Now, in Australia, it is 12. A similar trend of decreasing age of puberty is evident in males. Find out:
- **a** the reasons that researchers have suggested for the decreasing age of puberty in both females and males
- **b** the social problems that are arising through males and females reaching physical maturity at an earlier age.

9780170449090

12 REPRODUCTION PRODUCES OFFSPRING

UNIT 2 CONTENT

SCIENCE INQUIRY SKILLS

» design investigations, including the procedure(s) to be followed, the materials required, and the type and amount of primary and/or secondary data to be collected; conduct risk assessments; and consider research ethics, including animal ethics

SCIENCE AS A HUMAN ENDEAVOUR

» lifestyle choices, including diet, illicit drugs, alcohol and nicotine, may affect foetal development

SCIENCE UNDERSTANDING

Human reproduction

» for the establishment of a pregnancy, conception requires the union of viable sperm and ovum at the optimal time in the ovarian cycle

» the development of the embryo after implantation involves the differentiation of cells into three different germ layers that will eventually produce specific systems in the body and the placenta

» the stages of labour include birth, during which there are circulatory system changes in the child

Source: School Curriculum and Standards Authority, Government of Western Australia

9780170449090

12.1 FERTILISATION

In human reproduction, a sperm and an ovum are brought together at fertilisation. The resulting fertilised ovum, the zygote, develops into an embryo with many different types of cells.

Sexual intercourse

For fertilisation to occur, male sperm need to be brought into contact with an ovum produced by the female. The usual method by which this is brought about is **sexual intercourse**. For sexual intercourse to take place, and for sperm to be deposited in the vagina, the penis must become enlarged and firm, a condition referred to as an **erection**. An erection results from blood rushing into the spaces of the erectile tissue of the penis. Sexual excitation initiates this blood flow.

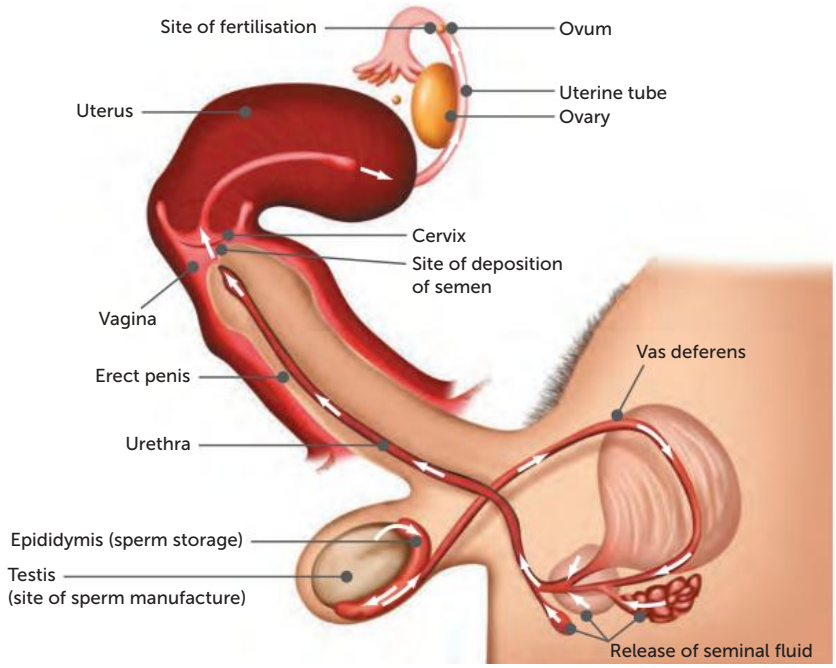


FIGURE 12.1 The pathway taken by sperm during sexual intercourse

When sexual stimulation of the penis within the vagina becomes sufficiently intense, rhythmic contractions of the epididymis, the vasa deferentia, the seminal vesicles and the prostate gland occur. The contractions propel the contents of the ducts and glands into the urethra and then out of the body. This process is called **ejaculation**. The ejaculated material consists of fluid, or **semen**, which contains sperm. Accompanying ejaculation is a rapid heartbeat, an increase in blood pressure and breathing rate, and intensely pleasurable sensations. These reactions constitute an **orgasm**.

An ejaculation normally expels about 3 mL of semen (about one teaspoonful), containing 250–300 million sperm. Besides the sperm, semen contains the secretions of the seminal vesicles, bulbo-urethral glands and prostate gland. The greatest contribution comes from the seminal vesicles, which produce a thick fluid containing nourishment for the sperm. Besides nourishment, semen provides the sperm with a fluid in which to swim, and neutralises the acid nature of the male urethra and female vagina. In addition, it contains enzymes that activate the sperm once ejaculation has taken place.

When the female is sexually stimulated, erectile tissue in the region of the vaginal opening fills with blood. This reduces the size of the vaginal opening and tends to increase the stimulation of the penis during sexual intercourse. Arousal also results in copious secretions of mucus by glands located around the cervix and in the region of the vaginal opening. These secretions lubricate the epithelial lining of the vagina, allowing for easy entry of the penis.

9780170449090

As sexual intercourse progresses, the external genitalia are rhythmically stimulated and, when sexual arousal reaches sufficient intensity, the female undergoes an orgasm, or **climax**. Female orgasm is somewhat like that of the male, with the exception that there is no ejaculation in the female. However, there may be an increase in the secretion of cervical mucus. A female does not need to reach an orgasm for fertilisation to occur, and it is still not known whether female orgasm helps fertilisation in any way.

Fertilisation

When the male ejaculates, the sperm are released in the vagina at the entrance to the uterus; a process called **insemination**. Once within the vagina, the sperm travel through the cervix and the body of the uterus into the uterine tubes. They quickly reach the upper portions of the uterine tubes, often within a few minutes. This rate is too fast to be due solely to the swimming motion of the sperm. It is thought that muscular contractions of the uterus and uterine tubes help to transport sperm through the female reproductive tract. At the same time, muscular contractions of the uterine tube, together with the beating action of cilia, transport the ova towards the uterus after ovulation.

Of the hundreds of millions of sperm deposited into the vagina during sexual intercourse, only a few thousand reach the uterine tubes. The death rate of sperm, called **sperm mortality**, is high, and is one reason why a large number of sperm are required if fertilisation is to occur. Fertilisation normally occurs in the uterine tubes when the ovum is about one-third of the way down the tube.

The secondary oocyte that is released at ovulation is at metaphase II. It is surrounded by two layers.

- The outer **corona radiata** consists of follicle cells held together by cementing materials that contain acid.
- The inner **zona pellucida** is a glycoprotein matrix surrounding the plasma membrane of the oocyte.

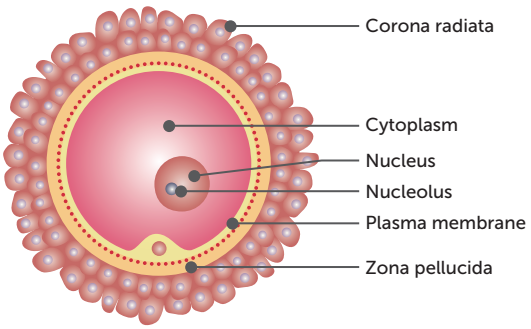


FIGURE 12.2 Structure of a secondary oocyte

The outer surface of the acrosome on the head of the sperm contains an enzyme. This enzyme is capable of breaking down the acid in the cementing material that holds the cells of the corona radiata together. However, the amount of enzyme contained in a single sperm is extremely small and ineffective. When several thousand sperm surround the oocyte, there is enough enzyme to loosen the cells of the corona, allowing one sperm to penetrate the corona radiata. This is another reason why a large number of sperm are required if fertilisation is to occur.

Once the sperm is through the corona radiata, it encounters the zona pellucida. This initiates the **acrosomal reaction**, causing digestive enzymes from the acrosome to be released. These enzymes break down the glycoprotein matrix of the zona pellucida, giving the sperm access to the plasma membrane of the oocyte. When the plasma membranes of the oocyte and spermatozoa fuse, the nucleus of the sperm enters the ovum.

The entrance of one sperm into the secondary oocyte stimulates the formation of a fertilisation membrane around the oocyte, which prevents the entrance of any more sperm. This ensures that only one haploid set of chromosomes joins the chromosomes of the oocyte.

9780170449090

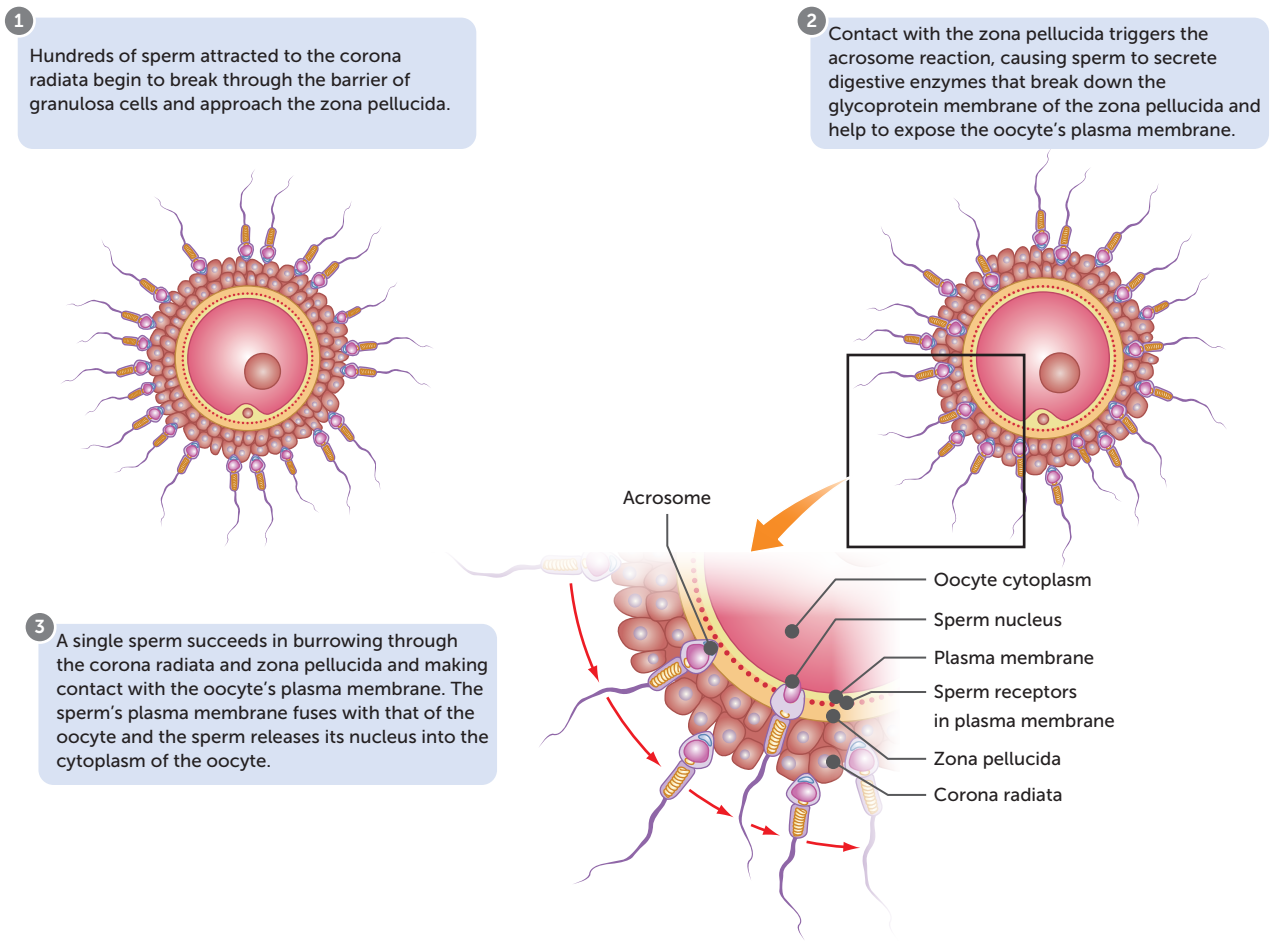


FIGURE 12.3 Process of fertilisation

Once the sperm has entered the oocyte the tail is absorbed, and the head begins to move through the cytoplasm in the form of a **male pronucleus** – the haploid nucleus of the sperm. The entrance of the sperm stimulates the secondary oocyte to complete the second meiotic division. The nucleus of the oocyte develops into a **female pronucleus** (a haploid nucleus of the oocyte), which fuses with the male pronucleus to form a single nucleus that now has the diploid number of chromosomes. Fertilisation is complete, and the fertilised oocyte is called a **zygote**.

Key concept

Fertilisation occurs when two haploid gametes, a sperm and an ovum, combine to form one diploid cell, the zygote.

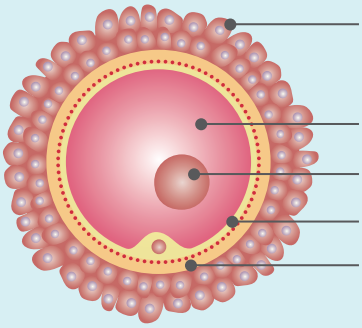
Fertilisation animation
Watch an animation showing the pathway of the sperm from insemination to fertilisation.

Questions 12.1

RECALL KNOWLEDGE

1. Define 'male pronucleus', 'acrosome', 'zygote', 'sperm mortality' and 'ejaculation'.
2. Describe the process that causes a penile erection.
3. List the components of semen.
4. List the processes that cause the movement of sperm through the female reproductive tract.

5 Label the plasma membrane, nucleus, cytoplasm, zona pellucida and corona radiata on the diagram below.



6 Describe the acrosomal reaction.

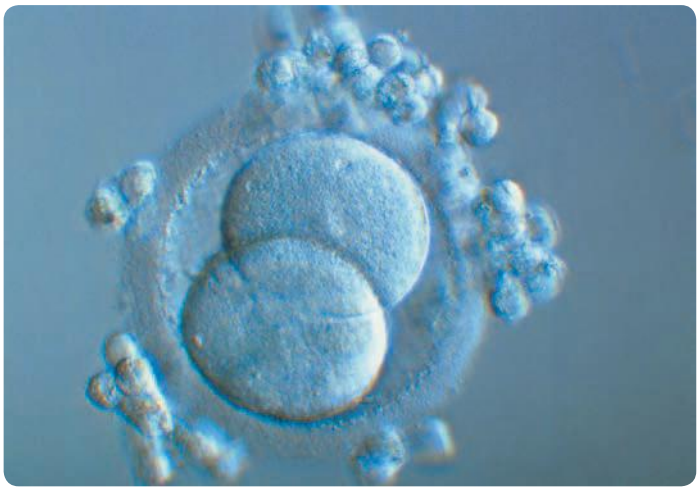
APPLY KNOWLEDGE

7 Explain why a large number of sperm are needed for the fertilisation of one ovum.

8 Use a flow diagram to demonstrate the processes from sexual intercourse to fertilisation.

12.2 EARLY EMBRYONIC DEVELOPMENT AND IMPLANTATION

The zygote, the single cell that results from the fertilisation of an ovum by a sperm, has the potential to grow into a new individual human. This means that more than 200 types of cells in the human body develop from that one initial cell.



Science Photo Library/Pascal Goetgheluck

FIGURE 12.4 Photograph of a human embryo at the two-cell stage. Here, the embryo is still surrounded by the gelatinous covering that was around the ovum

Blastocyst formation

After fertilisation, the zygote travels down the uterine (or Fallopian) tube and begins to divide by mitosis. The process of mitosis results in the formation of two cells exactly the same as the original parent cell. These two cells divide again by mitosis into four, then eight, then 16, and so on. By about six days after fertilisation, the original zygote has reached the uterus and has developed into a **blastocyst**. The blastocyst is a hollow ball of cells that surround a cavity filled with fluid. At one side of the cavity is a group of about 30 cells called the **inner cell mass** (sometimes known as the **embryoblast**). The inner cell mass is composed of stem cells that will differentiate into the different body cells to form the **embryo**.

9780170449090

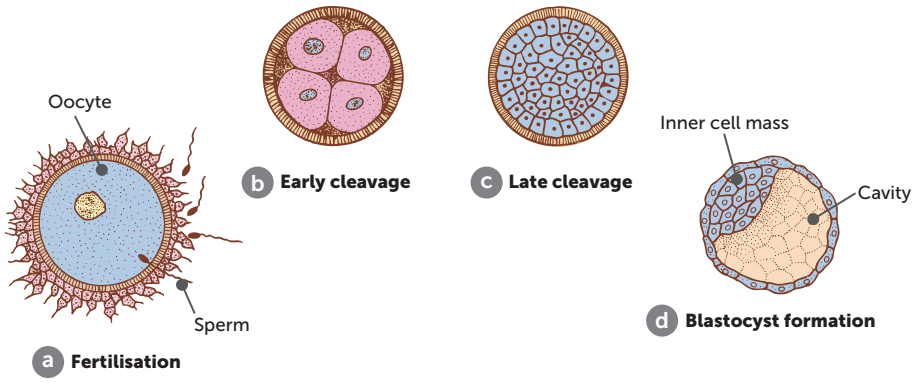


FIGURE 12.5 From fertilisation to the formation of a blastocyst

Implantation

The blastocyst remains free within the cavity of the uterus for two to three days, and then sinks into the soft endometrium (uterine lining) to become firmly attached to the wall of the uterus. This process is called **implantation**, and enables the blastocyst to gain nourishment for growth and development by absorbing nutrients from the glands and blood vessels of the uterine lining.

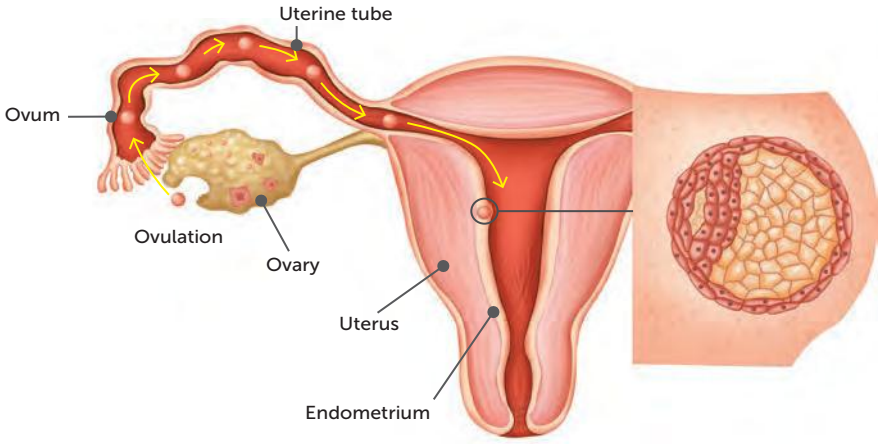


FIGURE 12.6 Implantation of a blastocyst

Hormone production

The continued development of the blastocyst depends on the endometrium being maintained. High levels of oestrogen and progesterone in the blood stop the endometrium breaking down, and so the menstrual cycle ceases. During the early stages of pregnancy, it is the corpus luteum that produces these hormones until the developing placenta can take over the role after approximately 8–12 weeks.

The first two months of pregnancy are referred to as the embryonic period; after the second month, the developing individual is called a **foetus**.

Pregnancy hormones
This website has a downloadable poster about the hormones produced during pregnancy.

Cell differentiation

The cells that make up the inner cell mass of the blastocyst are stem cells. As you learnt in Chapter 10, stem cells are very different from other cells because:

- they are not specialised for any particular role
- they are capable of repeated division by mitosis – a process called **proliferation**

- given the right conditions, they can differentiate into specialised cells. All the 200 or more types of cells that make up a mature human body develop from the stem cells of the inner cell mass.

Scientists are just beginning to understand the signals inside and outside cells that trigger stem cell differentiation. The internal signals are controlled by a cell's **genes**, while the external signals include chemicals secreted by other cells, and physical contact with neighbouring cells and certain molecules in the cell's immediate surroundings – its **microenvironment**.

After a sperm fertilises an oocyte, a zygote is formed, which then has the potential to develop into a complete embryo. At this point, the fertilised oocyte is a **totipotent stem cell**, which means that it has the potential to create any type of cell necessary for embryonic development, including the embryo itself, and all the membranes associated with embryonic development.

In the first few hours after fertilisation, the zygote undergoes several cell divisions that produce identical totipotent cells. Because these cells are still totipotent, any one of them has the potential to develop into an entire human being. In fact, identical twins are formed when two totipotent cells separate and develop into two genetically identical embryos.

The totipotent cells undergo several rounds of cell division. About five days after fertilisation, they begin to specialise and form a blastocyst. The outer layer of cells will eventually form the placenta and other tissues that are needed for the support and development of the foetus. The inner cell mass will form all the tissues of the human body; therefore, these are the cells that develop into the foetus. The cells of the inner cell mass are **pluripotent stem cells**. This means that they are able to give rise to many, but not all, cell types necessary for foetal development. For example, they are able to give rise to foetal tissues, but not placental tissue.

Each pluripotent cell then undergoes further specialisation into another type of stem cell – a **multipotent stem cell**. Multipotent stem cells give rise to cells that have a particular function; for example, blood stem cells give rise to red blood cells, white blood cells and platelets, whereas skin stem cells give rise to the different types of skin cells.

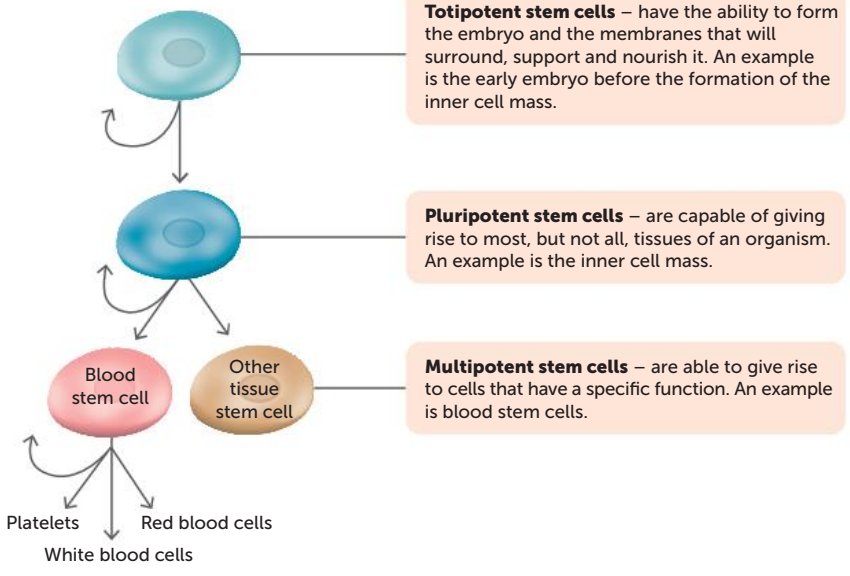


FIGURE 12.7 Process of cell differentiation

Stem cells
This website provides more information about stem cells.

Cell differentiation
Watch a video showing the zygote forming an embryo.

Key concept

A zygote develops from an unspecialised totipotent cell to a blastocyst, and then to an embryo and its membranes.

Primary germ layers

While the blastocyst is implanting in the lining of the uterus, during the third week of development, the inner cell mass undergoes changes as the cells change to multipotent. This process results in the formation of three layers of cells, the **primary germ layers**. These layers, called the **ectoderm**, **mesoderm** and **endoderm**, are the embryonic tissues that will differentiate into all the tissues and organs of the body. Table 12.1 lists the structures that are formed by the three primary germ layers.

Ectoderm

The ectoderm is the outermost germ layer. This will form the outer layers of the body, such as the skin, hair and mammary glands, as well as the nervous system.

Mesoderm

The mesoderm is the middle germ layer. The skeleton, muscles, connective tissue, heart, blood and urogenital tract form from the mesoderm. The mesoderm also allows the formation of the stomach and intestines.

Endoderm

The endoderm is the innermost germ layer. It forms the lining of the digestive system as well as the lungs and thyroid.

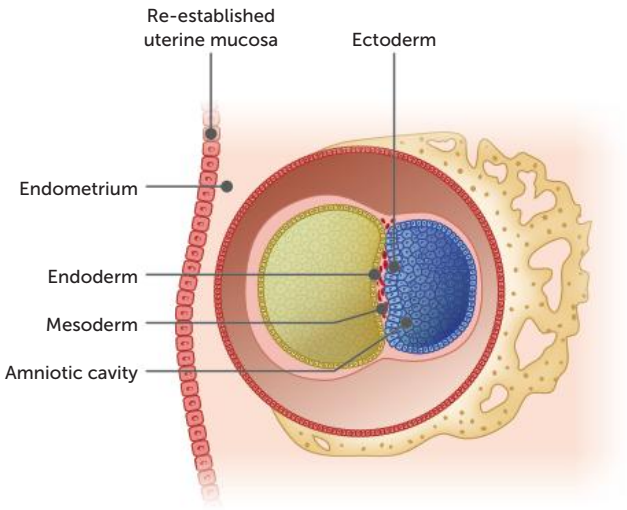


FIGURE 12.8 The primary germ layers: the ectoderm, mesoderm and endoderm

Key concept

The primary germ layers differentiate to form the specialised structures of the embryo.

TABLE 12.1 Structures formed by the three primary germ layers

ENDODERM	MESODERM	ECTODERM
Epithelium of alimentary canal and its glands (e.g. liver and pancreas)	Skeletal, smooth and cardiac muscles	Epidermis of skin
Epithelium of urinary bladder, urethra and gall bladder	Cartilage, bone, blood and other connective tissue	Hair, nails, glands of skin
Epithelium of pharynx, auditory canal, larynx, trachea, bronchi and lungs	Lymphoid tissue	Lens, cornea and muscles of the eye
Epithelium of tonsils, thyroid, parathyroid and thymus glands	Endothelium of blood vessels and lymphatics	Receptor cells of the sense organs
Epithelium of vagina and associated glands	Epithelium of the body cavity and joint cavities	Epithelium of mouth, nostrils, sinuses, glands of mouth, and anal canal
	Epithelium of kidneys and ureters	Enamel of the teeth
	Epithelium of ovaries, testes and reproductive tracts	Entire nervous system
	Epithelium of adrenal cortex	Anterior lobe of the pituitary gland
	Dermis of skin	Adrenal medulla

Embryonic membranes

Early in the embryonic period, four embryonic membranes form. These lie outside the embryo and serve to protect and nourish it as it develops.

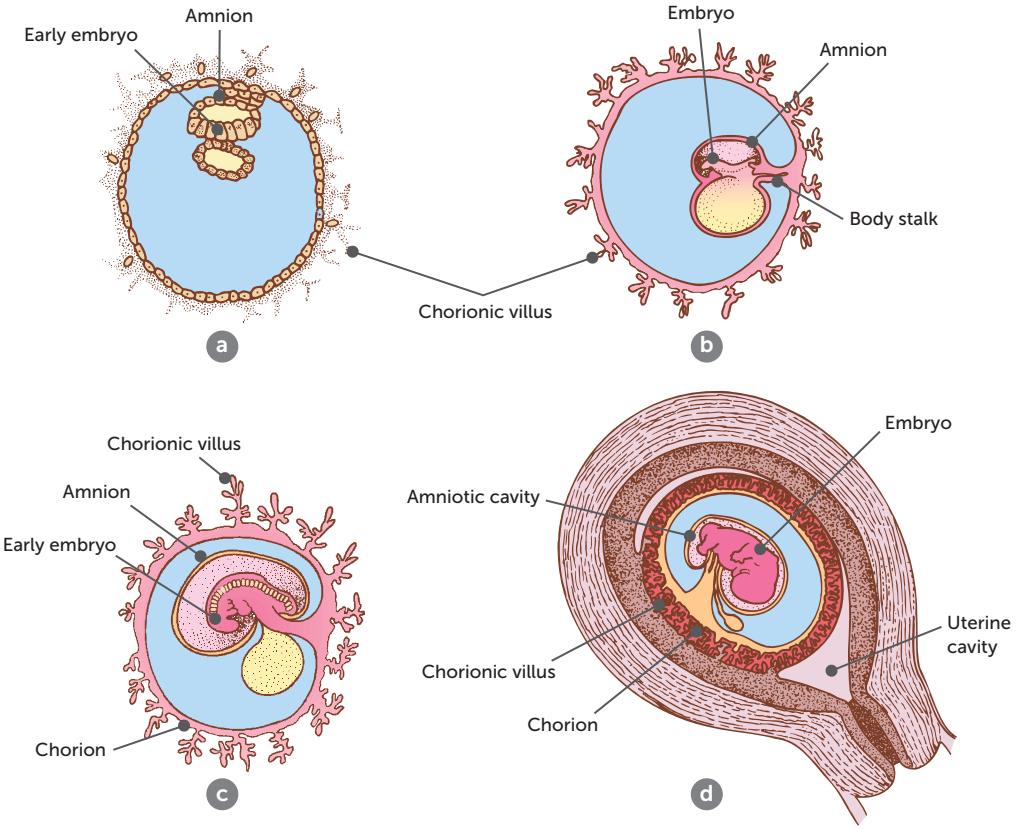


FIGURE 12.9 Development of the embryonic membranes

Amnion

The **amnion** is the first membrane to develop. By the eighth day after fertilisation, it surrounds the embryo, enclosing a cavity into which it secretes **amniotic fluid**. This fluid serves to protect the embryo against physical injury by acting as a shock absorber. It also helps to maintain a constant temperature and allows the developing embryo, and later the foetus, to move freely. The amnion expands as growth takes place. It usually ruptures just before childbirth, releasing the amniotic fluid, an event commonly referred to as 'breaking of the waters'.

Chorion

Another embryonic membrane is the **chorion**. It is formed from the outer cells of the blastocyst together with a layer of mesodermal cells. The chorion surrounds the embryo and the other three embryonic membranes. As the amnion enlarges, it fuses with the inner layer of the chorion. Eventually, the chorion becomes the main part of the foetal portion of the placenta.

Yolk sac and allantois

In addition to the chorion and the amnion, there are two other membranes – the yolk sac and the allantois. These are not as important in humans as they are in the development of many other animals; however, they do form the outer structure of the umbilical cord.

9780170449090

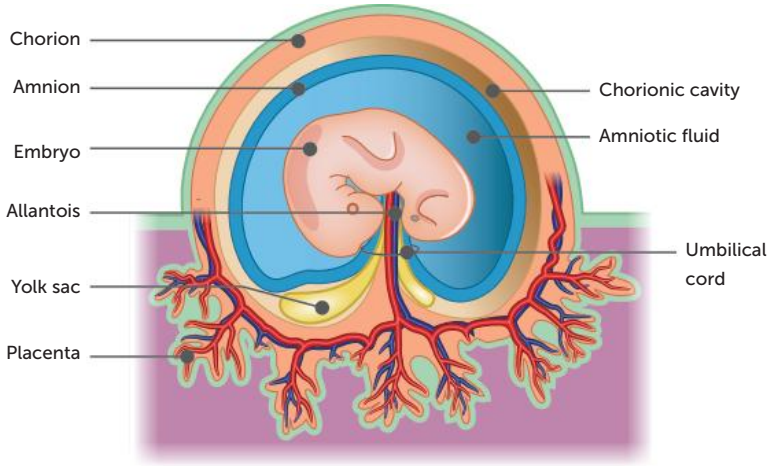


FIGURE 12.10 Embryonic membranes

Development of the placenta

The **placenta** is an organ that forms from both foetal and maternal tissues during the first three months of gestation, with the foetal portion developing from part of the chorion. The placenta supplies nutrients to, and removes wastes from, the foetus. It also serves as an endocrine organ, producing a number of the hormones necessary to maintain pregnancy. Table 12.2 lists its various functions.

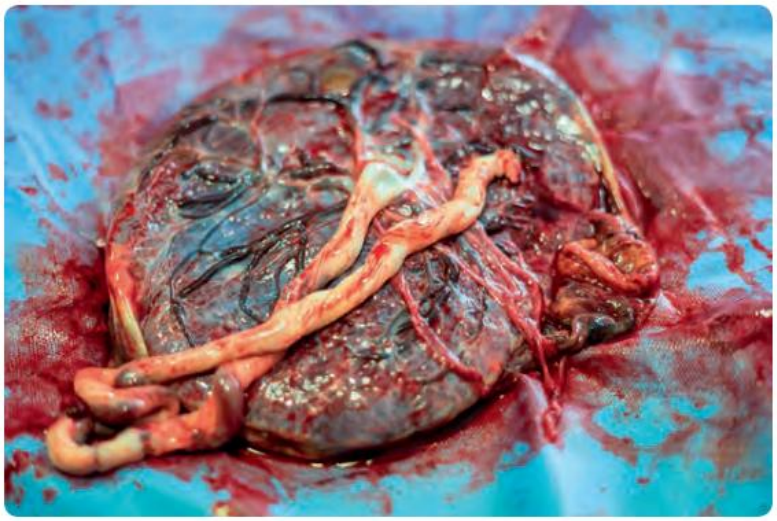


FIGURE 12.11 Human placenta

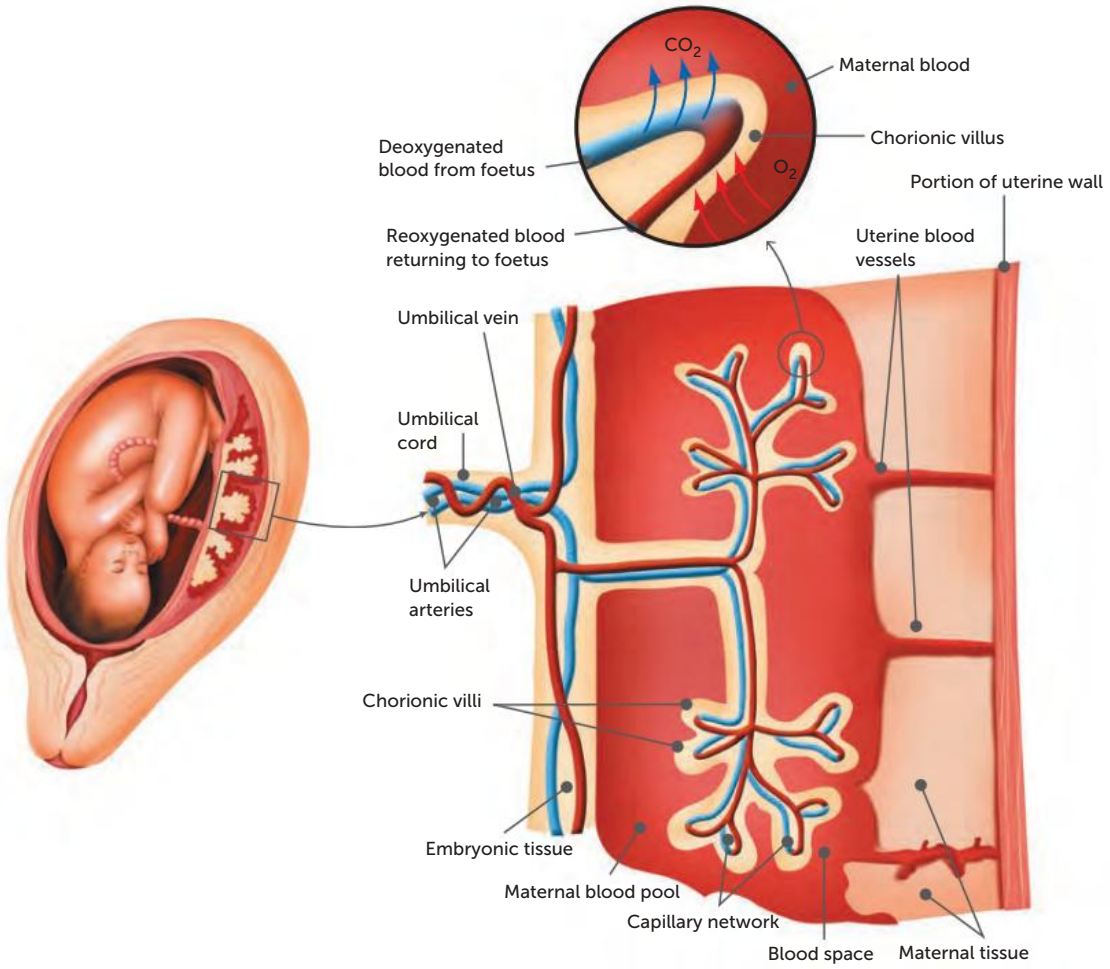
TABLE 12.2 Functions of the placenta

ROLE	FUNCTION
Endocrine	Secretes a number of hormones necessary for maintaining the pregnancy
Excretory	Transports nitrogenous wastes such as urea, uric acid, ammonia and creatinine from the foetal blood to the mother's blood supply for excretion by the mother's kidneys
Immune	Transports antibodies from the mother into the foetal blood supply so that the foetus has immunity to some infectious diseases
Nutritional	Transports nutrients such as glucose, amino acids, fatty acids, vitamins and minerals from the mother's blood to the foetal blood; stores some essential nutrients early in pregnancy and releases them later when the demand is greater
Respiratory	Transports oxygen from the mother to the foetus, and carbon dioxide from the foetus to the mother

The foetal part of the placenta begins to develop as the blastocyst is implanted in the endometrium. Small, branching, finger-like projections, called **chorionic villi,** develop from the chorion and contain numerous blood vessels. They grow into the endometrium, much like the root of a tree penetrating the soil. As the villi penetrate the endometrium, they become surrounded by pools of the mother's blood, which has collected in spaces within the endometrium. In this way, the villi are bathed in the mother's blood, but, the foetal and maternal blood do not normally mix because there are a few layers of cells separating the two blood supplies. However, the exchange of materials can take place by diffusion and active transport. Oxygen and nutrients from the mother's blood diffuse into the foetal blood, and wastes leave the foetus by diffusing into the maternal blood. The large number of villi that form provides an extensive surface area across which substances can pass. A fully developed placenta has an estimated surface area of about 16 m².

The placenta is attached to the foetus by the **umbilical cord**. Inside the umbilical cord are two **umbilical arteries** that carry blood to the capillaries of the chorionic villi. A single **umbilical vein** carries blood from the placenta, through the umbilical cord, back to the foetus. On the maternal side, blood from the mother enters the placenta through the uterine arteries, flows through the blood spaces where the exchange of substances occurs, and leaves again through the uterine veins.

FIGURE 12.12
Structure of the placenta and umbilical cord



Key concept

The placenta connects the embryo, or foetus, and the mother, allowing the mother to meet the needs of the baby.

Questions 12.2

RECALL KNOWLEDGE

1 Define 'zygote', 'stem cell', 'pluripotent' and 'primary germ layer'.
2 Draw a labelled diagram of a blastocyst.
3 List the key features of stem cells.
4 The cells produced by the initial cell divisions are totipotent. Suggest why this is important.
5 Name the primary germ layer that produces
   a hair
   b the lining of the lungs
   c bones
   d the brain
   e blood.
6 List the functions of the amniotic fluid.
7 Describe how blood flows from the
   a foetus to the placenta
   b placenta to the foetus
   c mother to the placenta
   d placenta to the mother.

APPLY KNOWLEDGE

8 Women who are trying to get pregnant may be prescribed progesterone to take after ovulation. Explain why this may increase the chances of pregnancy.
9 Prenatal check-ups monitor the health of the placenta. Suggest why this is important.
10 Use the position of the primary germ layers to justify the structures that they eventually form.

12.3 PREGNANCY

The period of pregnancy, the time that the embryo or foetus is carried in the uterus, is called **gestation**. During this period, the developing child grows to a length of about 50 cm and to an average weight of 3400 g. This growth and development takes about 280 days, measured from the beginning of the last menstrual period. The date of birth is normally predicted on the basis of the last menstrual period, because this date is known and the date of fertilisation is not.

From embryo to foetus

After one month of growth, the human embryo is just under 4 mm long. The most obvious feature at this stage is the development of muscle segments on either side of the tube that is to become the brain and spinal cord. These blocks of mesodermal tissue increase in number over time. By the end of the fourth week there are 30 pairs of them, representing the beginnings of the muscles and the vertebrae of the spinal column. In addition, the brain is beginning to form, a tail is evident, and the heart and liver are beginning to develop.

Additionally, the throat region contains a number of pharyngeal arches with clefts between them. As development proceeds, these arches will form the structural elements of the face and throat, and the pouches that develop in the clefts will give rise to the epithelial linings and glands associated with the throat.

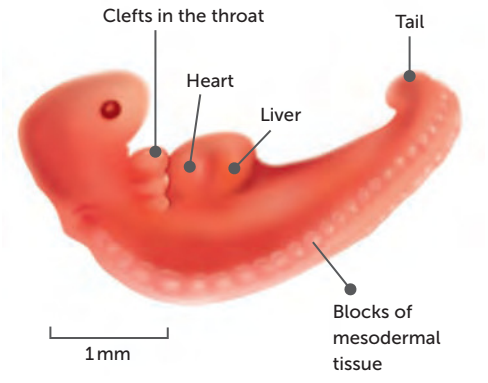


FIGURE 12.13 Embryo at the end of the fourth week

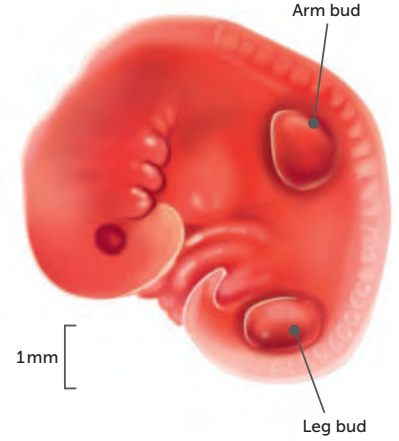


FIGURE 12.14 Embryo at the beginning of the fifth week

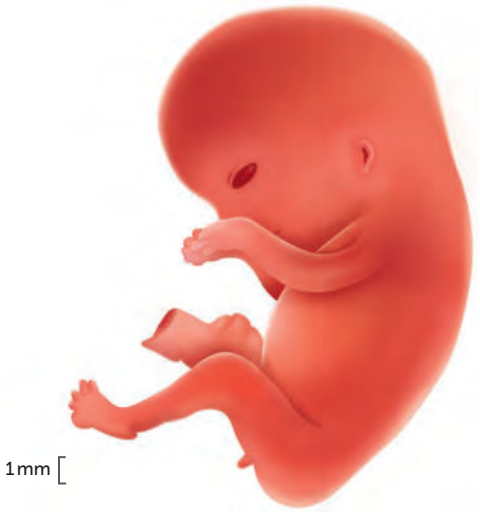


FIGURE 12.15
Embryo at the end of the seventh week

During the fifth week, the arm and leg buds start to appear. The arm buds are slightly more advanced, but both elongate rapidly from this time on.

By the end of the embryonic period (week 8), the embryo has a recognisably human form and all organs are present, although many are not fully functional. It has undergone considerable growth from a microscopic cell to being 3 cm in length from the top of its enlarged head to its buttocks, and weighing about 1 g. The head is almost half the size of the embryo and the eyes appear like slits, having moved from the sides of the head to be directed forward. The jaws are almost fully developed, as is the nose, and there are small earlobes. The arms and legs are well proportioned, and the hands are formed with distinctly human fingers. Similarly, the toes are well formed, and the external sexual organs are evident, so the embryo is clearly male or female.

By the end of the first two months of embryonic life, the general body form of the infant has developed, and the basic plan of the organ systems is in place. The developing baby is now known as a foetus.

Foetal development

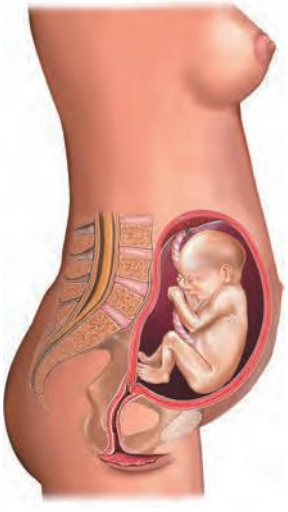
During the fourth month (the 16th week) of gestation, the uterus expands, and the woman's abdomen begins to bulge. The foetus grows rapidly during this month to about 18 cm long and 100 g in weight. Its posture is more erect, fingerprints appear and the foetus moves, stretching its arms and legs. The mother may begin to detect these movements. The heart beats strongly at 120–160 beats per minute, twice the rate of the mother's heart.

By week 20, the end of the fifth month, the foetus is about 25 cm long and weighs about 300 g. Foetal movements, such as kicking and turning, can now be felt clearly by the mother.

After 24 weeks of development, the mother is showing obvious signs of pregnancy. The foetus has grown to about 27–35 cm in length and weighs 565–680 g.

By the end of week 28, the foetus is about 38 cm in length, weighs over 1000 g, and moves around vigorously within the uterus. The brain has enlarged considerably, and its surface is now furrowed with developed functional areas. In the male, the testes usually descend into the scrotum during this period.

Embryo development
This website shows the development of an embryo at different stages.



Visuals Unlimited/Nucleus Medical Art

FIGURE 12.16 The 20-week foetus

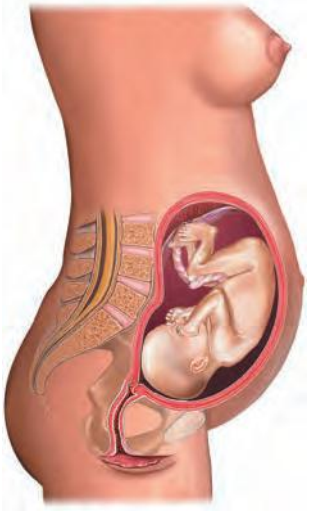


FIGURE 12.17 The 24-week foetus

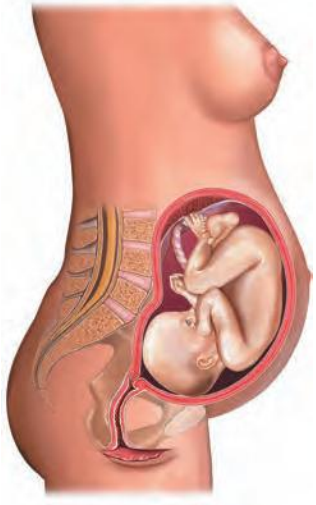


FIGURE 12.18 The 28-week foetus

9780170449090

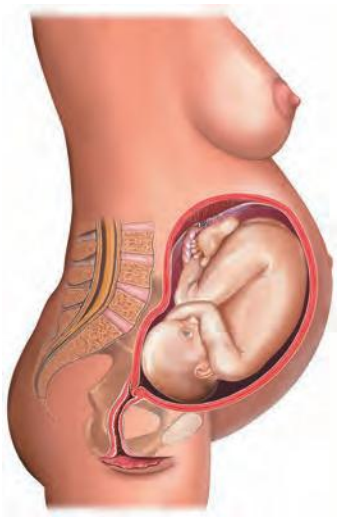


FIGURE 12.19 The 32-week foetus

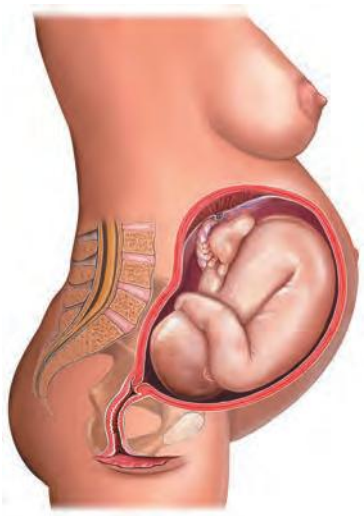


FIGURE 12.20 The 36-week foetus

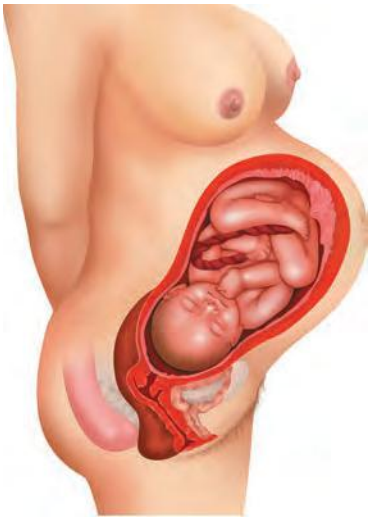


FIGURE 12.21 The foetus at full term

By the end of week 32, the foetus is 41–45 cm in length and weighs 1800–2200 g.

By week 36, the foetus is approximately 2700 g in weight and 46–48 cm in length. The circulatory system is fully developed, ready for birth. However, the digestive system still needs time to mature.

By week 40, the pregnancy is at full term. The foetal activity evident in earlier weeks is now diminished, as the foetus occupies all the available space within the uterus – it simply has no room to move. By this stage the foetus is about 50 cm in length and weighs about 3400 g. Boys are usually about 100 g heavier than girls. However, the birth weight of a baby can vary considerably, from as little as 2500 g to as much as 4500 g. Because of the growth of the body, the head is now smaller in proportion to the size of the body. The nose is well formed.

Shortly before birth, the foetus changes its position in the uterus and comes to lie with its head resting inside the curved shape of the pelvis. The movements of the foetus in this position are even more restricted than before. Growth of the foetus at this stage is also very slow, as the placenta begins to fail and becomes more fibrous.

During the later stages of pregnancy, antibodies from the mother diffuse across the placenta into the baby's blood. These give the newborn child temporary immunity against diseases to which the mother is immune. After about six months the effects of the antibodies gradually decrease as the child begins to develop its own immune responses.

From fertilisation to birth
This website provides pictures and information about the zygote, embryo and foetus.

Activity 12.1
Summarising development

Key concept

During pregnancy the body systems of the embryo, and then the foetus, develop so that they are able to meet the needs of the baby at birth.

TABLE 12.3 Stages of foetal development

AGE (MONTHS)	FOETAL DEVELOPMENT
3	Forelimbs well developed; eyelids closed; outer ear completed; bone marrow formed; blood cells formed in bone marrow; sex distinguishable
4	Arms and hands fully shaped; skeleton completed; exercising of muscles evident; ears stand out from head
5	Fine hair covers body; gripping reflexes are developed; increased growth
6	Respiratory movements; digestive glands begin to function; tooth buds evident; eyebrows and eyelashes

TABLE 12.3 (*Continued*)

AGE (MONTHS)	FOETAL DEVELOPMENT
7	Period of greatest growth; all systems functional except respiratory system
8	Accumulation of fat beneath skin; growth slowed
9	Eyes open; nose well formed; sucking and grasping reflexes apparent; fine body hair is shed

The pregnant mother

The most obvious changes to the pregnant woman are those associated with her growing abdomen: the abdomen bulges as a result of the growth of the uterus. Figure 12.22 shows the outline of the uterus at various stages after implantation.

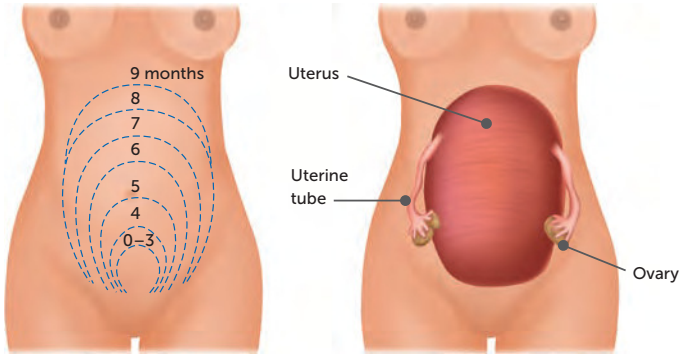


FIGURE 12.22 Size and position of the uterus at various stages of pregnancy

Not all the increase in the size of the abdomen is due to the uterus. Some is due to other internal organs, such as the stomach, liver and intestines, being forced upwards and outwards.

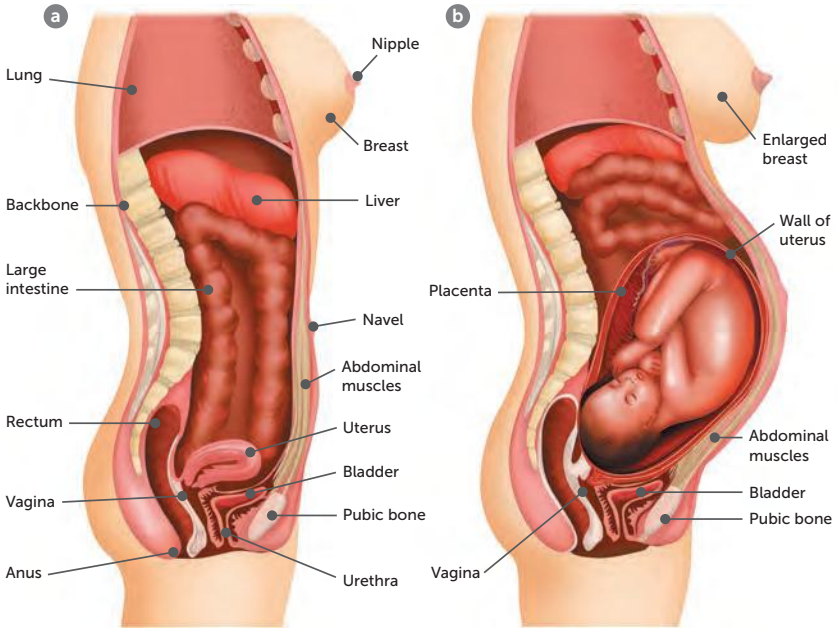


FIGURE 12.23 Internal organs of woman who **a** is not pregnant, and **b** is pregnant

Another obvious change during the course of pregnancy is the enlargement of the breasts. The hormones of pregnancy result in the development of the milk-secreting tissues, which leads to an increase in size.

Pregnancy also affects the mother in less obvious ways.

- There is an increase in the size of the heart and in blood volume. This is to cater for the extra blood that is flowing through the placenta.
- The greater blood volume results in an increased blood flow to the kidneys and, therefore, increased urine production. Additionally, during the first three months of pregnancy, the

expanding uterus presses on the bladder so that it feels as if it is filled with urine. As the uterus grows, it moves up the pelvic cavity, releasing this pressure. Then, during the last stages of pregnancy, the foetus presses on the bladder again.

- The emotional state of the mother may be affected due to the changes in hormonal balance and as a result of natural fears accompanying pregnancy. The mother may be concerned about her child's development, the problems that may occur at the time of birth, and the effect the newborn child will have on the rest of the family. Many of these factors are beyond the control of the pregnant woman and so support and reassurance from family and friends are very important in maintaining a positive outlook.

Key concept

A mother's body changes during pregnancy to accommodate the growing foetus.

Questions 12.3

RECALL KNOWLEDGE

1 Construct a timeline showing the length and mass of the embryo from 4 weeks until 40 weeks.
2 At what age does an embryo become a foetus?
3 State the age when each of the following occurs:
   a eyes form as slits
   b kicking and turning can be felt clearly
   c the testes descend into the scrotum
   d fingerprints appear
   e the brain begins to develop
   f arm buds form
   g the brain is well developed and has furrows.
4 List the reasons for increased urination during pregnancy.

APPLY KNOWLEDGE

5 Explain the difference between pregnancy and gestation.
6 Babies born prematurely often need to be put on a respirator to breathe for them. Suggest the reason for this.
7 Explain why heavily pregnant women often prefer to eat smaller meals than normal.

12.4 CHANGES DURING BIRTH

The process by which the foetus is expelled from the mother's body at the end of gestation is called birth, or **parturition**. Parturition is preceded by a sequence of events commonly called **labour**.

The birth process

Prior to labour

In preparation for labour, several hormonal changes occur. These changes cause the ligaments of the pelvis to soften, making them more pliable for childbirth. The hormonal changes also increase the response of the uterus to stimuli and strengthen contraction of its muscles.

Before labour begins, the foetus has probably settled with its head in the mother's pelvis. The cervix has softened, shortened in length, and is likely to have begun to open a little. The foetus is usually facing the woman's right or left hip bone, with its knees drawn up to its abdomen and its legs crossed. In this position it takes up as little room as possible. One side of its head is usually pressed against the mother's bladder, the other against her bowel.

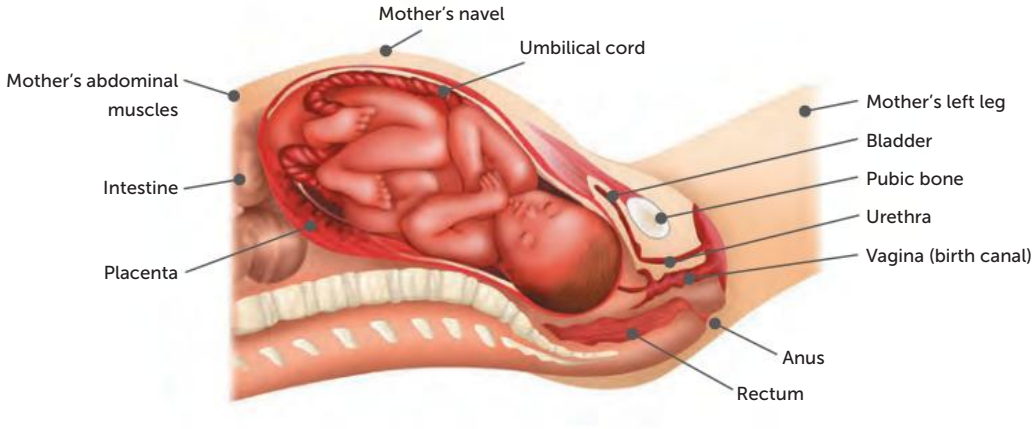


FIGURE 12.24 The foetus just before birth

First stage of labour

During the final three months of gestation, the uterus undergoes weak, irregular contractions. These contractions gradually become stronger and more frequent during the last weeks of pregnancy. Eventually the contractions become strong and occur about every 30 minutes. This is the beginning of the birth process and the contractions are known as **labour pains**.

The first stage of labour, the **dilation of the cervix**, is the time from the onset of labour to the complete dilation (or opening) of the cervix. Although variable in length, it lasts an average of eight to nine hours if it is the birth of the woman's first child and about four hours for the birth of subsequent children.

Waves of contraction travel from the upper part of the uterus downward towards the cervix. These waves are similar to the peristalsis that occurs in the alimentary canal to push food along. With each contraction, the muscle fibres making up the uterus shorten a small amount, pulling on the cervix. This pull on the cervix shortens it so that it no longer projects down into the vagina. At the same time, the cervix is opened. This cervical dilation allows the foetus to move more deeply into the pelvis.

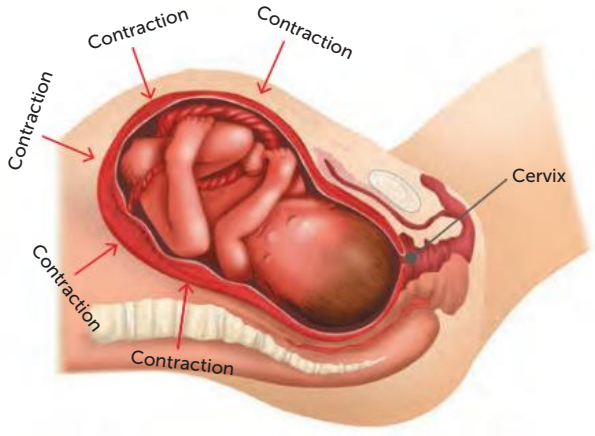


FIGURE 12.25 The first stage of labour

As the contractions become more frequent and stronger, the head of the foetus is pushed more forcefully against the slowly dilating cervix. Eventually the cervix is completely dilated (usually to about 10 cm), and the uterus, cervix and vagina form a single, curved passage. This passage, termed the **birth canal**, is the route through which the foetus will pass, aided by the contractions of the uterus and voluntary contractions of the abdominal muscles of the mother. Complete dilation of the cervix marks the end of the first stage of labour.

Second stage of labour

The second stage of labour involves the delivery of the foetus and is often called the **stage of expulsion**. It frequently begins with the bursting of the membrane surrounding the foetus and a gush of fluid from the vagina. This may occur much earlier in labour or, occasionally, may not occur until the foetus is ready to be born. In most cases, however, it occurs at the beginning of the second stage of labour.

9780170449090

The second stage, from full dilation of the cervix to birth, lasts from 20 minutes to 2 hours. As the foetus moves through the fully dilated cervix, its head stretches the vagina. This distension of the vagina stimulates the woman to contract her abdominal muscles. These contractions, together with the contractions of the uterus, push the foetus through the vagina. As this occurs, the baby's head turns to face towards the mother's back.

With each contraction, the head advances a small amount. Between contractions it retreats a little, but overall the head gradually moves through the external opening of the vagina. As this occurs, more and more of the head becomes visible. During this time, the mother is working very hard (she is really in labour!). Her pulse rate increases, and she usually begins to sweat from the effort required. Between contractions the mother tries to rest a little, to gain strength for the next effort. Eventually the head stretches the vaginal entrance and the tissues between it and the anus. This tissue becomes tightly stretched over the foetus's head as it is forced into the world. Once the head has emerged, it turns sideways again to face the mother's hips. This rotation allows the shoulders and the rest of the body to move more easily through the birth canal.

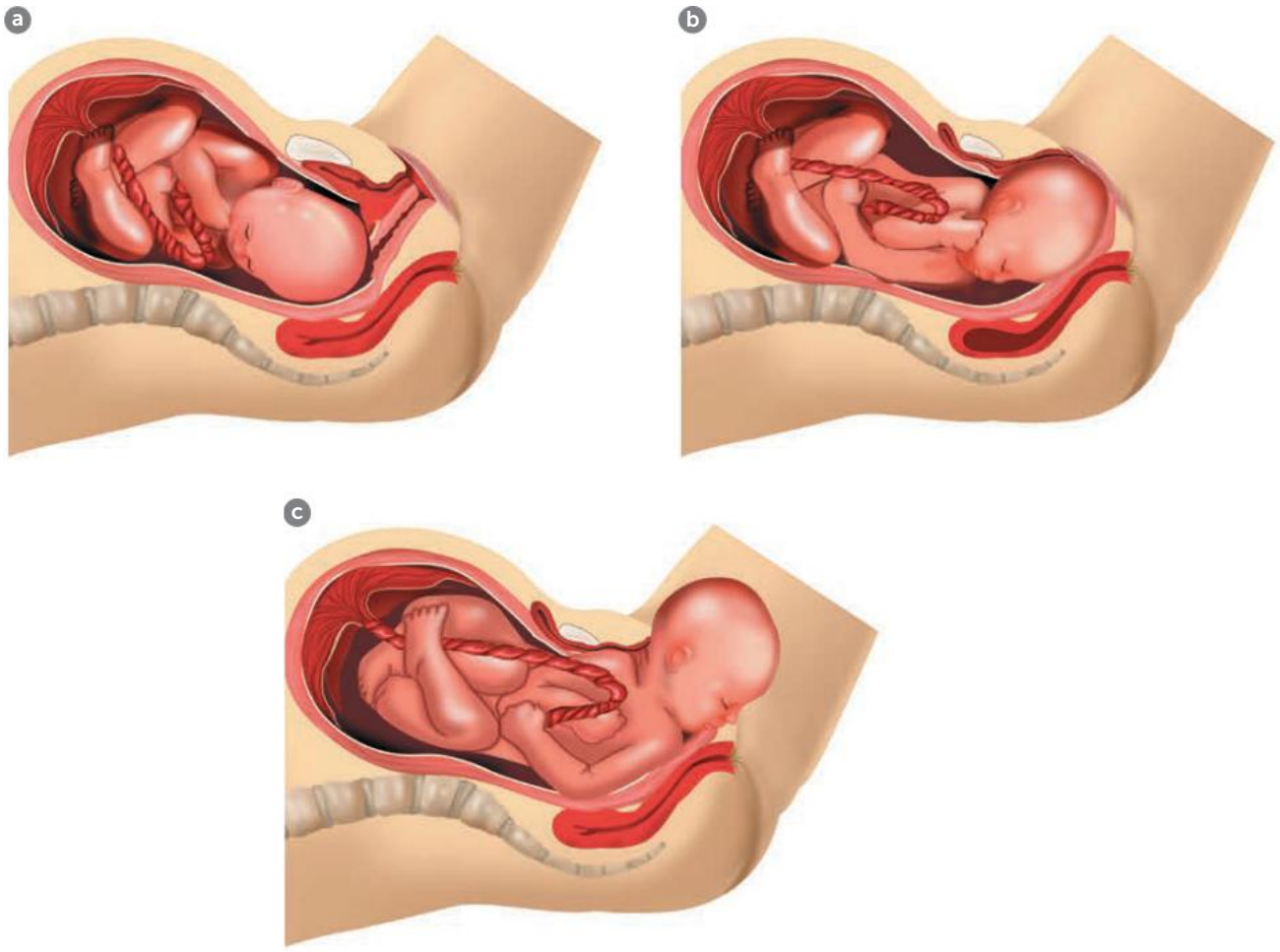


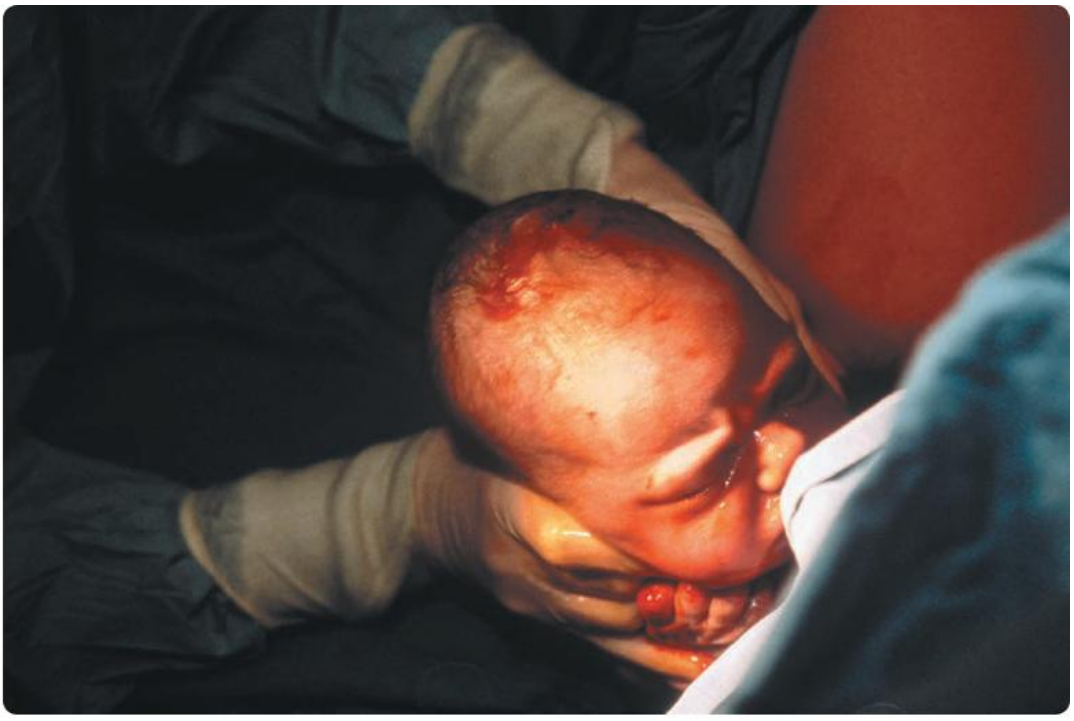
FIGURE 12.26 a Early in the second stage of labour: the baby's head is starting to turn so that it faces towards its mother's back and the amnion has ruptured; **b** Late in the second stage: the baby's head appears at the entrance to the vagina and its shoulders are turning to fit into the bones of the pelvis; at this stage, the face is turned completely towards the mother's back; **c** The baby's head emerges from the vagina

As the foetus passes through the birth canal, the pressure on the head may push it out of shape. The underlying brain is not damaged, as the bones of the skull are pliable and separated by joints that allow some degree of overlap. The head resumes its normal shape a few days after birth.

9780170449090

The head of the foetus is downward in over 90% of births. This position allows the head to be delivered first and to act as a wedge to force open the cervix and the vagina. The head-down position also allows the foetus to begin breathing even before it is completely free of the birth canal.

FIGURE 12.27 Birth of a baby



Science Source/Southern Illinois University

Third stage of labour

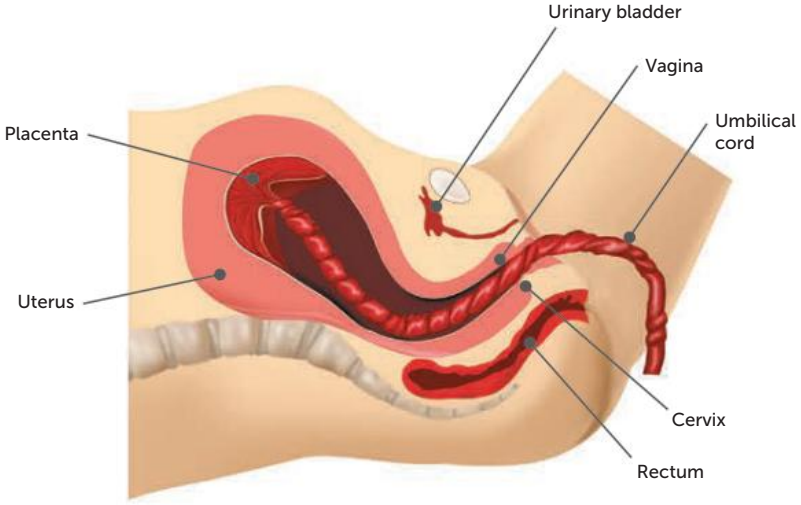
Once born, the baby begins to breathe with its own lungs, even though it is still connected to the placenta by the umbilical cord. The amnion, chorion and placenta are still inside the uterus at this stage. The umbilical cord is clamped, tied in two places, and then cut between the ties. The arteries and vein within the umbilical cord contract, either before or immediately after they are cut. After a few days, the stump of the cord dries up and falls away. The navel, or **umbilicus**, is all that remains.

WS

12.1 Stages of labour

The uterus continues to contract, and about five minutes after delivery the placenta, other membranes and the remains of the umbilical cord are expelled. Together these are called the **afterbirth**. Little blood is lost during this stage as the placental blood vessels

FIGURE 12.28 Third stage of labour: the afterbirth is expelled from the uterus



constrict and contractions of the uterus squeeze shut the uterine vessels that supply blood to the placenta. Blood clots then form to stop all leakage of blood. With such a large area of exposed tissue, infection can occur. In the past this was quite common and made childbirth hazardous, with many women dying from infection of the uterus. Today, with strict standards of cleanliness and the availability of antibiotics, women seldom die from infection following childbirth.

Key concept

During the three stages of the birth process, the mother's body undergoes changes to allow the baby, and the afterbirth, to be expelled.

Changes in the baby at birth

During development, the embryo, and later the foetus, is totally dependent on the mother for all its needs. The mother supplies the foetus with oxygen and nutrients, eliminates carbon dioxide and other wastes, and protects it against changes in temperature, shocks and many disease-causing organisms. At birth, this all changes: the infant has to become self-supporting.

Before birth, the lungs of the foetus do not function. Therefore, it obtains its oxygen from the placenta. In the same way, the foetus gets its nutrients from the placenta rather than from its own alimentary canal. These important differences between the foetus and the baby after birth mean that the circulation must change when the baby is born.

Ductus venosus

Before birth, foetal blood is carried to and from the placenta by blood vessels in the umbilical cord. The baby's blood is carried to the placenta in two umbilical arteries. As it circulates through the placenta, carbon dioxide and other wastes are exchanged for oxygen and nutrients. The blood then returns to the foetus via the umbilical vein. Some of the blood returning to the foetus flows through the liver and into the inferior vena cava (the main vein taking blood to the heart from the lower body). The remainder, approximately 30%, bypasses the liver by flowing through a vessel called the **ductus venosus**, and then into the inferior vena cava. The fact that much of the blood does not pass through the liver causes no problems at this stage, as the mother's liver is serving the needs of the foetus.

Ductus arteriosus and foramen ovale

Blood returning to the foetal heart enters the right atrium. From there it can follow several pathways:

- Blood may flow into the right ventricle and then to the lungs in the usual way. However, the lungs are collapsed and not functioning at this stage, so they offer considerable resistance to blood flow and little blood reaches the lungs.
- Most of the blood from the right ventricle flows through the ductus arteriosus to the aorta. The **ductus arteriosus** is a vessel that bypasses the lung, allowing blood in the pulmonary artery to flow directly into the aorta.
- Blood in the right atrium of the heart may flow directly into the left atrium through an oval opening between the two chambers. This opening is called the **foramen ovale**. The foramen ovale is located so that most of the blood entering the right atrium goes through it. This is beneficial, as the blood coming from the placenta is highly oxygenated and can flow to the developing foetal tissues via the aorta very quickly.

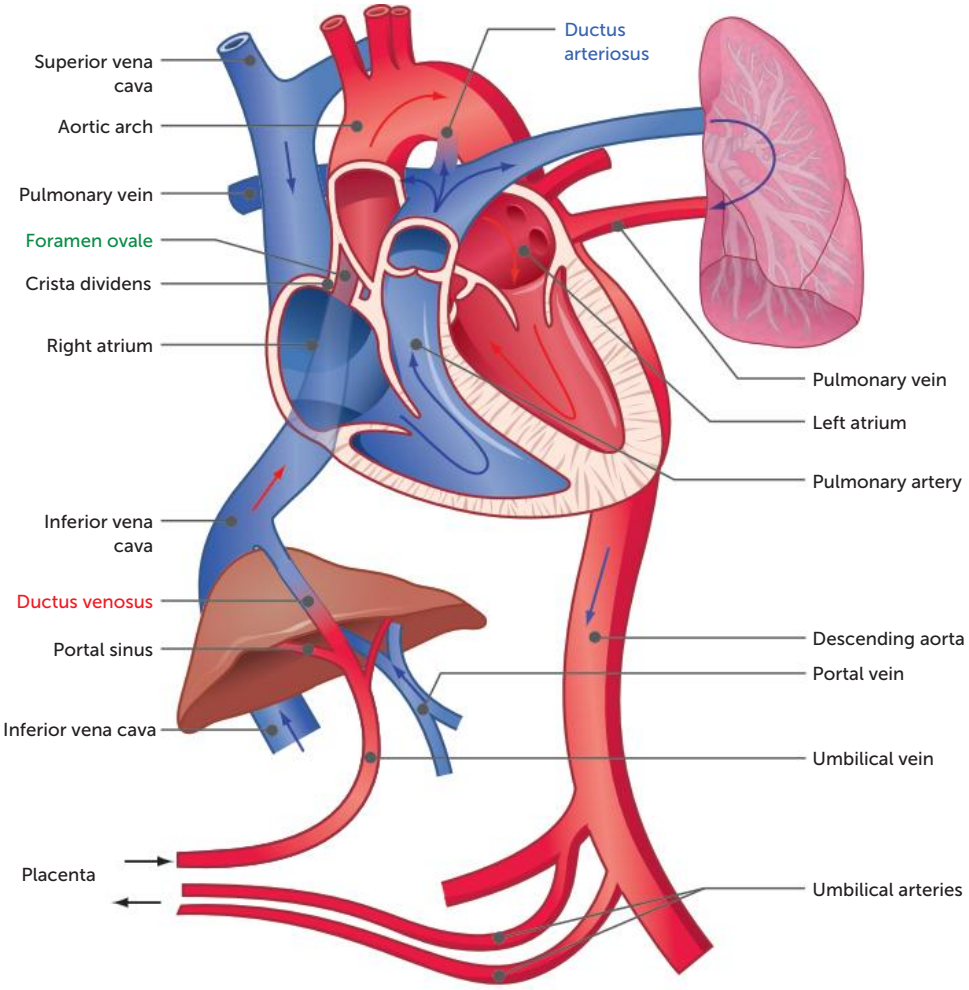


FIGURE 12.29
Foetal circulation showing the ductus venosus (red), ductus arteriosus (blue) and foramen ovale (green)

Changes at birth

At birth, the newborn can no longer depend on the placenta for food and oxygen. The lungs and liver must now become fully functional, and for this to occur blood must flow through them. Therefore, it is important that the ductus venosus, ductus arteriosus and foramen ovale close.

The first breath of life is usually triggered by the shock of birth. If not, the traditional slap on the baby's bottom provides a stimulus that initiates breathing. Failing this, the clamping of the umbilical vessels allows the level of carbon dioxide in the baby's blood to rise. This stimulates the respiratory centre in the brain and as a result the lungs begin to function. As the lungs expand, they no longer offer the same resistance to blood flow, so blood flow through the ductus arteriosus begins to decrease. A few weeks after the birth, all that is left of the ductus arteriosus is some fibrous tissue.

As larger amounts of blood return to the heart from the lungs, the pressure in the left atrium increases. This increased pressure forces the flap of the foramen ovale against the wall of the atrium, closing off the opening. Eventually, the foramen ovale becomes permanently closed.

With the cutting of the umbilical cord, blood no longer flows through the umbilical vessels or the ductus venosus. As no blood is being carried through it, the ductus venosus gradually constricts until it is permanently closed off. The bypass around the liver is then lost and all blood in the blood vessel to the liver must pass through the liver.

If the foramen ovale fails to close, the baby may be born with a 'hole in the heart'. The first indication of this is at birth, as the baby has a bluish colour due to insufficient oxygen in the blood. Because the foramen ovale is still open, not enough blood flows through the lungs. Surgery can be performed to close the foramen ovale and provide normal circulation.

Changes to circulation
This website has more information about the circulation of a foetus and the changes that occur after birth.

Following its birth, the baby breathes rapidly, at about 45 breaths per minute, for the first two weeks. The breathing rate then gradually slows. Similarly, the heart rate of the newborn is high: it may be from 125 to 130 beats per minute, often going as high as 180 beats per minute at times of excitement. The rate is high because more oxygen is needed for increased muscular activity and to keep the baby warm in the cooler environment outside the uterus. For the same reason, the number of red blood cells increases, to carry extra oxygen to the tissues. The white blood cell count, on the other hand, is very high at birth but decreases rapidly by the seventh day.

Key concept

During pregnancy, the baby's circulatory system has a foramen ovale, ductus venosus and ductus arteriosus, as the mother is providing the needs of the baby. After birth, the baby needs to meet its own needs and so these structures resolve.

Questions 12.4

RECALL KNOWLEDGE

1. What term is used to describe the birth of a baby?
2. Use a flow chart to summarise the birth process.
3. State the average length of the first stage of labour for a woman having her:
   a. second child
   b. first child
   c. fourth child.
4. Describe the birth canal.

APPLY KNOWLEDGE

5. Explain the advantage of having a foramen ovale.
6. Describe the changes that cause the closure of the ductus arteriosus following birth.
7. Explain the importance of the position of the foetus just before birth.
8. On TV, it is common for a woman giving birth to say that they 'have to push'. Discuss whether this is a realistic statement.
9. The ductus venosus could be compared to a bypass highway: a road that goes around busy areas of traffic. Discuss whether or not this analogy is accurate.

12.5 MAINTAINING A HEALTHY PREGNANCY

Throughout pregnancy, all women hope that the developing child will be born healthy. To help achieve this goal, a woman wishing to have a child needs to ensure that she takes care of her own body so that if she does become pregnant, her developing child will then have the best chance for a healthy life. Women wishing to become pregnant, or who are already pregnant, can make lifestyle choices that will reduce the risk of damage to the baby.

Supplying the foetus's requirements

While the embryo, and later the foetus, develops in the uterus, changes occur in the mother's body to allow her to adjust to the needs of pregnancy. These changes start very early in pregnancy, and most of them take place ahead of the demands that the foetus will place on its mother for oxygen, nutrients and waste removal. During the embryonic period, when the organ systems are developing, the woman easily supplies the oxygen and nutrients that the embryo requires. In the later weeks of foetal development, when large amounts of oxygen and nutrients are needed, adjustments are required in the functioning of the woman's body. Her own functions slow down, allowing nutrients to

stay in the blood for a longer period of time. This enables them to be more easily diffused across the placenta for use by the foetus.

Slowing of body functioning has some disadvantages for the mother. As her alimentary canal is less active, her stomach empties more slowly, and constipation is therefore common. As the concentration of nutrients in the bloodstream is higher, more tend to be filtered out by the kidneys and are lost in the urine. This loss is easily compensated for by a well-balanced diet.

For the developing foetus to obtain the nutrients it requires, a large quantity of blood needs to flow through the placenta. This is met by a gradual increase in the volume of the mother's blood and a faster rate of circulation through her blood vessels. This results from an increase in both the rate at which the heart beats and the amount of blood pumped with each beat. By the end of pregnancy, the mother's blood volume will have increased by 40%.

Diet

Diet is an important aspect of prenatal care. However, its importance should not be overstressed because, provided the diet is balanced, its influence on the birth weight and survival of the baby is minimal.

The average pregnant woman needs an increase in energy intake of about 850 kJ per day, especially in the second half of her pregnancy. Pregnancy also requires an increased protein intake to ensure that the developing foetus is adequately supplied. The diet should contain at least 65 g of protein each day. Other important dietary requirements for a pregnant woman are as follows:

- Folic acid (folate) is essential for normal cell division and for the manufacture of protein. Lack of folic acid before and during pregnancy can also contribute to spina bifida and other neural tube defects. In this condition, the bony arch of the vertebrae around the spinal cord does not develop. Mothers can help to protect their babies from problems like spina bifida by increasing their folic acid intake at least one month before pregnancy and for the first three months of pregnancy. Foods rich in folic acid are wholegrain breads and cereals, green leafy vegetables and legumes.
- Adequate amounts of calcium are necessary for normal bone growth, as well as for teeth, heart, nerve and muscle development.
- In areas where fluoride is not added to the drinking water, fluoride tablets after the 20th week of pregnancy will help to protect the foetus from future dental problems.
- Vitamin A is required for the normal growth of cells and, as little is stored in the body, a steady intake is necessary. During pregnancy, the demand increases considerably, especially in the last 10 weeks, and the level of this vitamin in the woman's blood tends to fall. However, if the pregnant woman has a balanced diet, with good quantities of green and yellow vegetables, there should be no need for concern.

Listeria infection, or **listeriosis**, is a very mild illness caused by eating food contaminated with the bacterium *Listeria monocytogenes*. Infection in pregnant women has the potential to cause miscarriages or stillbirths. To guard against listeria infection, pregnant women should eat food that has been freshly prepared or cooked. Foods to avoid are salads from salad bars, pre-packaged salads, soft cheeses, pâté, and raw or smoked seafoods. Pregnant women should also carefully read any warnings on food labels to ensure there is no risk to the developing child.

Weight gain

Weight gain can be a problem for some women during pregnancy. The mother will obviously gain weight as her pregnancy progresses, especially due to the foetus, the placenta and the amniotic fluid. Increases in blood volume and the size of the breasts and uterus also contribute to weight gain. The hormonal changes involved in pregnancy promote the conversion of energy to fat and the retention of water in the body, both contributing to an increase in weight. It is best if the mother can keep her weight gain to about 0.5 kg a week during the second half of the pregnancy. Excessive weight gained during pregnancy is very hard to lose after the child is born, particularly if the mother is not breastfeeding.

9780170449090

Exercise

If a pregnant woman is used to regular exercise, she should maintain her exercise program. Women who are not used to exercise should not suddenly start a program just because they are pregnant, although walking regularly is a good habit to establish and maintain.

A number of studies have found that exercise is usually safe, and that women who have a good exercise program:

- are more likely to carry their babies to full term than women who do not regularly exercise
- are better able to maintain their stamina during labour
- regain their pre-pregnancy body health and fitness more quickly after the birth.

Activity 12.2
Investigating pregnancy and exercise

Exposure to teratogens

A **teratogenic agent** (or **teratogen**) is one that causes physical defects in the developing embryo. There is a wide range of potential teratogens, including some hormones, antibiotics, oral anticoagulants, anticonvulsants, anti-tumour drugs, thyroid drugs, thalidomide, LSD (lysergic acid diethylamide) and marijuana. The range of actions and the effects of teratogens vary significantly and depend on the substance, the dose and the stage of development.

Teratogens are usually identified after an increased prevalence of a certain birth defect. For example, the increased prevalence of cerebral palsy in babies born to mothers living around Minamata Bay in Japan led to researchers identifying methyl mercury as the offending agent. A local factory was discharging this chemical into the water in the bay and contaminating the fish. After pregnant women ate the contaminated fish, the methyl mercury passed across the placenta and affected the developing foetus. Affected babies were born suffering convulsions, intellectual disabilities and general brain damage.

Alcohol

Alcohol was a suspected teratogen for hundreds of years. It is only relatively recently that a relationship between maternal alcohol intake and characteristic malformations in the foetus has been recognised. **Foetal alcohol syndrome (FAS)** is the term used to describe the effects of foetal exposure to alcohol. It appears that 1 in every 1000 births may be affected by FAS. While it is unlikely that an alcoholic drink now and then causes harm to the mother's developing baby, health authorities advise women who are pregnant, or planning pregnancy, not to drink alcohol. Excessive alcohol intake, especially 'binge drinking' early in pregnancy, definitely has a marked effect on the child. The most obvious effect is a lower than normal birth weight. Other symptoms include slow growth before and after birth, a small head, irregularities of the face such as narrow eye slits and sunken nasal bridge, defects of the heart and other organs, malformed arms and legs, and intellectual disabilities. Besides such physical abnormalities, the child may have behavioural problems, such as hyperactivity, extreme nervousness and a poor attention span.

Smoking

Smoking during pregnancy has an adverse effect on the developing foetus. The birth weight of babies born to women who smoke during pregnancy is significantly lower than that of babies born to women who do not smoke. In addition, there is evidence of an increased risk of miscarriage. Children of mothers who smoke and breastfeed their babies are more likely to suffer from gastrointestinal problems than other children. Children of smoking mothers also have a higher incidence of respiratory problems, including bronchitis and pneumonia, during the first year of life. There is a strong association between smoking during pregnancy and sudden infant death syndrome (SIDS). If the mother resumes smoking after giving birth, there is still an increased risk of SIDS in the child.

SIDS
Use this website to find out more about SIDS.

Chemicals

Besides alcohol and the ingredients of cigarette smoke, many other chemicals are known to be teratogenic agents. One is **thalidomide**, a chemical that was originally developed for use in sleeping pills. It was also found to be effective in the prevention of morning sickness during the first months of pregnancy. Two years after it went on sale in 1958, a sharp rise in the incidence of certain limb malformations was noticed. The malformations were of a type that is normally very rare, and this was a major factor in the discovery of the teratogenic action of thalidomide. If the malformations had been less conspicuous, they may not have aroused medical attention so quickly. Two doctors, Widukind Lenz in Germany and William McBride in Sydney, were responsible for linking these malformations to thalidomide. By the time thalidomide was taken off the market in November 1961, an estimated 7000 babies had been affected.

Getty Images/Paul Fievez

FIGURE 12.30
Many children of women who took the drug thalidomide during pregnancy in the 1950s had malformed limbs and other disabilities

Thalidomide is a good example of where the time of exposure to the teratogen determines the type of defect. Thalidomide acts on the embryo between the 28th and 42nd days of development, a time when the future limbs are forming. After 10 days of development the limbs start to appear, first as microscopic buds, then gradually developing into readily recognisable forms by the 42nd day. The arms are the first to develop, followed by the legs, which may be the reason why thalidomide affects the arms more frequently than the legs.

Other drugs

Illegal drugs such as heroin and LSD, as well as many medicinal drugs, can cause damage to the foetus when taken during pregnancy. Most doctors are extremely careful when prescribing drugs to women of child-bearing age, as many drugs cause the greatest harm early in pregnancy, when a woman may not even know she is pregnant. Any sexually active woman who is not using contraception should be very careful about using drugs of any kind without first seeking medical advice. The labels on most medicinal products now have clear warnings about possible side effects, and a pregnant woman should read such labels carefully to ensure there is no risk to her developing child.

Rubella

Rubella is a viral infection that was frequently contracted by school-aged children. It is a fairly mild disease, although highly infectious. However, if contracted by a pregnant woman it can have disastrous consequences for the child, who may be born deaf, blind or with heart malformations. In 1941, an Australian doctor, Norman Gregg, first made the connection between rubella and a high risk of birth abnormalities.

The rubella virus tends to grow in tissues that are just forming. Nine out of 10 babies infected during the first 10 weeks of pregnancy have a major problem such as deafness, blindness, heart defects or brain damage. The risk of damage decreases as the pregnancy progresses: the risk is 61% if infection occurs in the fourth month, and 10% towards the end of the pregnancy.

One of the vaccines recommended in the National Immunisation Program Schedule for children from birth to four years is MMR – measles, mumps, rubella. The vaccine, injected under the skin at 12 months and again at 18 months, gives lifelong protection against rubella. MMR vaccination began in Australia in 1989, and during the 1990s the number of rubella cases in Australia fell by more than 80% due to high rates of immunisation.

Key concept

The health of the mother and baby is affected by the mother's lifestyle and choices during pregnancy.

Questions 12.5

RECALL KNOWLEDGE

1. Define 'teratogen' and 'foetal alcohol syndrome'.
2. By what percentage does the mother's blood volume increase during pregnancy?
3. List three nutrients that pregnant women require in a higher level than non-pregnant women.
4. Explain why females who are trying to get pregnant should take folic acid supplements.
5. State five lifestyle choices that pregnant women should make.
6. List the reasons that women gain weight during pregnancy.
7. State the most common consequence of drinking alcohol during pregnancy.
8. Describe the effects of thalidomide on the embryo.

APPLY KNOWLEDGE

9. Explain why pregnant women are likely to experience constipation, especially late in the pregnancy.
10. During pregnancy, a woman's resting heart rate may rise to 90 beats per minute. Explain why this is higher than a non-pregnant woman's heart rate.
11. Explain why pregnant women should not eat soft cheeses such as camembert.

9780170449090

CHAPTER 12 ACTIVITIES

ACTIVITY 12.1 Summarising development

During pregnancy, the offspring changes from a single cell to a complex organism. Use a method of your choice to create a video of this process. Some possibilities are:

Your pictures

- Draw diagrams and take photos of each.
- Find images from the Internet. (Include your sources in a bibliography.)
- Draw cartoon images and take photos of each.

Your video

- Label your photos or images and use them in a program such as iMovie. Add headings and a voice-over to your images to explain the process.
- Use your photos or images in a program such as PowerPoint or Keynote. Add headings or subheading pages, labels, animations and voice-overs to explain the process. Export the presentation as a video.

ACTIVITY 12.2 Investigating pregnancy and exercise

Some observations have indicated that mothers who exercise during pregnancy seem more likely to avoid a premature delivery than those who do little or no exercise. During this task, you will investigate this relationship.

What to do

1. Identify the dependent and independent variables in this investigation.
2. Propose a hypothesis that states a relationship between the two variables.
3. Use Google Forms, SurveyMonkey, Microsoft Forms or a similar program to create a survey that could be given to mothers after the birth of their babies. The questions should be designed so that the answers will either support or disprove your hypothesis. In making up your questions, keep the following points in mind.
   - Make sure the questions are concise and clear. The respondents should not have to interpret what the question means.
   - Try to frame questions that require a yes/no answer or one response to several choices (multiple choice). If respondents are required to write a sentence answer, you may then have to interpret the answer.
   - Keep your questionnaire as brief as possible, but make sure the answers will give you enough information to reach a conclusion about your hypothesis.
4. If you know a mother with a baby, you may like to try out your questionnaire on her. If necessary, you could then modify the questionnaire.

Studying your observations

1. What pattern of answers to your questions would support your hypothesis?
2. How would you go about conducting your survey? In particular, how would you select participants?
3. How many participants would be necessary to enable you to decide whether your hypothesis was supported or disproved?

9780170449090

CHAPTER 12 SUMMARY

- Male and female gametes are brought together by sexual intercourse.
- During sexual intercourse, an erect penis releases semen into the vagina through an ejaculation. This process is called insemination.
- Sperm travel via the cervix into the uterus before moving up the uterine tubes.
- For a sperm to fertilise the oocyte, it must penetrate the corona radiata and zona pellucida. Enzymes on the acrosomes of a large number of sperm break down the cement holding the corona radiata together. An acrosomal reaction then releases digestive enzymes, allowing a single sperm to move through the zona pellucida and fuse with the plasma membrane of the oocyte.
- When the male pronucleus and female pronucleus join, they form a diploid cell called a zygote.
- The zygote undergoes mitosis, forming a hollow, fluid-filled ball called a blastocyst, which implants itself in the wall of the uterus.
- The inner cell mass of the blastocyst divides and differentiates to form the embryo.
- The zygote is a totipotent stem cell. The inner cell mass is made of pluripotent cells, which form multipotent stem cells after some specialisation.
- The blastocyst divides into the ectoderm, which forms the outer layers of the body; the mesoderm, which forms the skeleton, muscles and connective tissue; and the endoderm, which forms the lining of the digestive tract and lungs.
- Embryonic membranes protect and nourish the embryo. The amnion surrounds the embryo and contains the amniotic fluid. The chorion surrounds the embryo and the other embryonic membranes. It becomes the main foetal portion of the placenta.
- The placenta connects the foetus and mother to provide nutrients, remove wastes, and produce necessary hormones.
- The umbilical cord has two umbilical arteries and an umbilical vein that carry blood between placenta and foetus.
- During pregnancy, the embryo grows in size and develops muscles, bones and organs. Limbs then start to grow and the external sexual organs are evident.
- By the end of the first two months, the embryo is termed a foetus and has the general form of an infant.
- During its development, the foetus starts moving, becomes more erect and increases in size.
- During pregnancy, the mother's abdomen increases in size (reflecting the size of the foetus), her breasts enlarge, and blood volume increases.
- Prior to birth, the foetus moves so that its head rests in the pelvis; hormones soften the pelvic ligaments in the mother.
- During the first stage of labour, uterine contractions gradually increase in strength and the cervix dilates until it forms the birth canal with the uterus, cervix and vagina.
- During the second stage of labour, the membrane bursts and contractions push the foetus through the birth canal.
- In the final stage, the placenta, other membranes and the remains of the umbilical cord are expelled.
- Following birth, the baby's blood must flow through the lungs and liver. Therefore, the structures that bypass these organs during pregnancy – the ductus arteriosus, foramen ovale and ductus venosus – must close.
- Mothers can look after themselves and their baby during pregnancy by eating a well-balanced diet that includes folic acid, calcium, fluoride and vitamin A; avoiding foods that may cause listeriosis; controlling weight gain; and maintaining an exercise routine.
- Some chemicals, such as hormones, antibiotics, thalidomide, marijuana, alcohol and cigarettes, can affect the development of the foetus. These are known as teratogens.

9780170449090

CHAPTER 12 GLOSSARY

Acrosomal reaction The release of enzymes from the acrosome when the spermatozoa fuses with the ovum

Afterbirth The placenta, and the remains of the umbilical cord, amnion and chorion; it is expelled shortly after the birth of a baby

Amnion The membrane that forms a cavity around the embryo; it secretes a fluid (amniotic fluid) into the cavity to protect the embryo

Amniotic fluid The fluid contained within the amnion; it protects the embryo against injury

Birth canal The passage formed by the uterus, the dilated cervix and the vagina, through which the foetus travels at birth

Blastocyst A hollow ball of cells formed during early embryonic development

Chorion One of the embryonic membranes that eventually forms part of the placenta

Chorionic villi Finger-like projections that develop from the outer layer of cells of the early embryo; they grow to form part of the placenta; singular: chorionic villus

Climax The moment of intense pleasure during sexual intercourse

Corona radiata The innermost layer of cells surrounding the egg (or ovum)

Dilation of the cervix The first stage of labour, from the beginning of labour to the complete opening of the cervix

Ductus arteriosus The foetal blood vessel that enables blood in the pulmonary artery to bypass the lungs and flow directly into the aorta

Ductus venosus The foetal blood vessel that enables blood to bypass the liver

Ectoderm The outer tissue layer of the embryo; it gives rise to the outer covering of the body

Ejaculation Muscular contractions that propel semen from the penis

Embryo The early stage of development of an organism; in humans, from fertilisation to the end of the eighth week of pregnancy

Embryoblast *see* inner cell mass

Endoderm The inner tissue layer of the embryo; it gives rise to the lining of the digestive tract and the glands associated with it, as well as to the respiratory tract and parts of the excretory and reproductive systems

Erection The penis in a state of sexual excitation

Female pronucleus *see* pronucleus

Foetal alcohol syndrome (FAS) Abnormalities in a newborn baby due to the mother's excessive alcohol consumption during pregnancy; frequently results in lower than normal birth weight, a small head and slow growth

Foetus The developing individual after the second month of pregnancy

Foramen ovale An opening between the atria of the foetal heart; it allows blood to flow directly from the right atrium into the left atrium

Gene The factor that determines a hereditary characteristic; part of a chromosome

Gestation The period of development of an organism in the uterus; the time between conception and birth

Implantation The process whereby an embryo sinks into the lining of the uterus

Inner cell mass The group of cells in the blastocyst that develop into the embryo; also called the embryoblast

Insemination The deposition of sperm within the vagina

Labour The sequence of events that precede birth and result in the expulsion of the foetus through the vagina

Labour pains Contractions of the uterus that indicate the beginning of labour

Listeriosis An illness caused by eating food contaminated with listeria bacteria

Male pronucleus *see* pronucleus

Mesoderm The middle tissue layer of an embryo; it gives rise to the muscles, connective tissues and the alimentary canal

9780170449090

Microenvironment The immediate surroundings of a cell

Multipotent stem cell Stem cells that are able to give rise to a limited number of other cell types; for example, blood stem cells will give rise to red blood cells, white blood cells and platelets

Orgasm The climax of sexual intercourse

Parturition The process of birth

Placenta The organ that supplies nutrients to, and removes wastes from, the foetus; it also produces a number of hormones, including oestrogen and progesterone

Pluripotent stem cell Stem cells that are able to give rise to many, but not all, of the cell types necessary for foetal development

Primary germ layer The embryonic tissues from which all tissues and organs of the body will develop: the ectoderm, endoderm and mesoderm

Proliferation The process by which cells replicate themselves

Pronucleus The nucleus of the ovum (female: pronucleus) and of the sperm (male: pronucleus) that fuse at fertilisation

Rubella A viral infection that, if contracted by a pregnant woman, can have serious consequences for her unborn child; the child may be born deaf, blind or with heart malformations

Semen The liquid that nourishes and aids the transport of sperm; also called seminal fluid

Sexual intercourse The sex act during which gametes are passed from the male to the female

Sperm mortality The death rate of sperm

Stage of expulsion The second stage of labour, from full dilation of the cervix to birth

Teratogen An agent that causes physical defects in a developing foetus

Teratogenic agent *see* teratogen

Thalidomide A teratogen that causes babies to be born with limb malformations

Totipotent stem cell A stem cell able to create any of the types of cell necessary for embryonic development

Umbilical arteries Two arteries within the umbilical cord

Umbilical cord The cord that attaches the foetus to the placenta

Umbilical vein A single vein within the umbilical cord that carries blood from the placenta to the foetus

Umbilicus The small scar on the abdomen that marks the former attachment of the umbilical cord to the foetus; commonly called the navel

Zona pellucida The thick glycoprotein membrane surrounding the plasma membrane of the ovum

Zygote The fertilised ovum from which a new individual develops

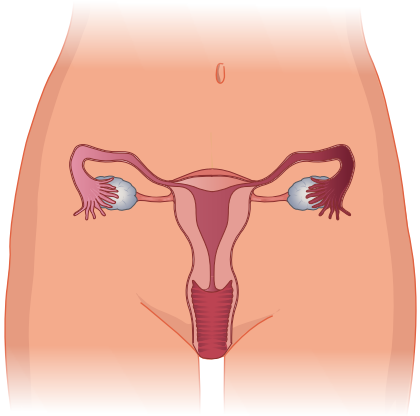
9780170449090

CHAPTER 12 REVIEW QUESTIONS

Recall

1 a Define 'fertilisation'.
   b Describe the events that take place in humans so that fertilisation can be achieved.
   c Name the structure where fertilisation normally occurs.
   d What is a zygote?

2 On the diagram below, clearly identify where:
   a sperm are deposited
   b fertilisation takes place
   c implantation occurs.



3 Describe the process of implantation.

4 a Draw a labelled diagram of a blastocyst.
   b At what stage of embryonic development does a blastocyst occur?
   c What type of stem cells exist in the blastocyst?

5 a Name the three embryonic germ layers.
   b Give two examples of tissues that develop from each of the germ layers.

6 Describe the placenta, including the embryonic tissues it develops from and the functions it performs.

7 Describe the main features of the eight-week-old embryo.

8 Draw a diagram to show how blood from the embryo/foetus gets to and from the placenta.

9 List the changes that occur in the mother during pregnancy.

10 a Make a list of factors that a pregnant woman should consider in regards to her diet.
   b Women gain weight during pregnancy. In addition to the growing foetus, what else contributes to the weight gain?

11 List the benefits of exercise for a pregnant woman.

12 a Define the term 'teratogen' (or 'teratogenic agent').
   b List examples of teratogenic agents.

13 Distinguish between gestation and parturition.

14 Briefly describe the events that take place during the:
   a first stage of labour
   b second stage of labour
   c third stage of labour.

15 Briefly describe the stimuli that normally trigger the newborn's first breath.

Explain

16 a Explain the need for the production of very large numbers of sperm in order for fertilisation to take place.
   b Besides sperm, what other components make up the semen?

17 Distinguish between proliferation and differentiation.

18 Compare and contrast an embryo and a foetus.

19 Explain how amniotic fluid helps the development of the foetus.

20 To meet the requirements of the developing foetus, a large quantity of blood needs to flow through the

9780170449090

placenta. Describe the changes in the mother's body that make it possible for a lot of blood to flow through the placenta.

21 Describe the changes that take place in the baby's circulatory system at birth. In doing so, ensure that you explain clearly the role of the ductus arteriosus, foramen ovale, ductus venosus and umbilical blood vessels in the foetal circulation.

Apply

22 Can a woman become pregnant the first time she has sexual intercourse? Explain.

23 Explain how the human male and female reproductive organs are arranged so that sperm can be transferred from the body of the male to the female for fertilisation to occur.

24 Why should a pregnant woman read the labels of prepared foods and medicinal products?

25 Explain why menstruation does not take place during pregnancy.

26 What changes take place in the uterus, cervix and vagina to allow them to function as a 'birth canal'?

Extend

27 Animals that breed in water, such as crayfish, fish and frogs, have no penis or vagina. Explain the advantages of these organs for a mammal, such as a human.

28 Explain how a blastocyst can consist of many more cells than a zygote, yet be only slightly larger in size.

29 Since the introduction of vaccination for rubella, the incidence of the disease has dropped, significantly reducing birth defects due to this disease. Find out what other vaccination programs could help reduce harm to the developing foetus.

30 Describe the survival advantage of having the umbilical vessels constricting before they are cut.

31 Australian doctors, in general, prefer women to have their babies in a hospital. Discuss the advantages and disadvantages of this approach compared with a homebirth.

32 If the foramen ovale fails to close, a baby may be born with a 'hole in the heart'. Use references to determine how often this birth defect occurs. What reasons are suggested for the failure of the foramen ovale to close? How soon after birth do doctors operate to rectify this situation?

33 A newborn infant has a rapid breathing rate and heart rate. Find out why these rates are high and for how long they stay high. What is the survival advantage for the infant?

34 One way of checking a male's fertility is to do a sperm count. Conduct research to find out:

- **a** what 'sperm count' means and what is considered a normal sperm count
- **b** whether there is anything a male can do to increase his sperm count
- **c** whether it is possible for a male with a low sperm count to father children.

35 In some pregnancies, and for a variety of reasons, the doctor looking after the woman may decide to induce labour. Reasons for this may include high blood pressure, bleeding, incompatibility of blood groups between foetus and mother, or a pregnancy that has gone on for more than 42 weeks. However, for most women, labour starts naturally.

- **a** Find out about the initiation of labour under natural circumstances.
- **b** Does medical intervention to induce labour mimic the natural process? Describe the similarities and differences.
- **c** How long do such induction techniques take to be effective?

36 There is some concern in society regarding stem cells. Conduct research into the source of stem cells and their uses. Use this to discuss the reasons for people's concerns and whether they are valid.

9780170449090

13 REDUCING THE CHANCE OF PREGNANCY AND STIs

UNIT 2 CONTENT

SCIENCE INQUIRY SKILLS

» interpret a range of scientific and media texts, and evaluate processes, claims and conclusions by considering the quality of available evidence; and use reasoning to construct scientific arguments

SCIENCE AS A HUMAN ENDEAVOUR

» greater understanding of the menstrual cycle, conception and implantation has produced improved methods of the establishment of a pregnancy, along with advancements in contraceptive methods; both have ethical considerations

SCIENCE UNDERSTANDING

Human reproduction

» contraception methods that reduce the probability of the union of gametes or implantation all have limitations, risks and benefits, and include methods that:
  – use steroid hormones
  – use physical barriers between gametes
  – use chemical spermicides
  – use sterilisation (tubal ligation, vasectomy)
  – function after coitus (emergency contraceptive pill and intrauterine devices [IUDs])

» sexually transmitted infections (STIs), diseases transmitted through unprotected sex or genital contact, can be prevented through safe sex methods; early detection and treatment of infection are important and, if left untreated, STIs can lead to serious health consequences

Source: School Curriculum and Standards Authority, Government of Western Australia

9780170449090

Pregnancy is a possible outcome of sexual activity. Another possible consequence is acquiring a sexually transmitted infection. Advances in human biological sciences have enabled us to understand the reproductive system to facilitate safe sex practices. This allows individuals to plan pregnancies and reduce the chance of infections.

13.1 CONTRACEPTION

It is only in relatively recent times that people have been able to plan a family. Although some of the family planning techniques described in this chapter have been used for centuries, they were notoriously unreliable. Modern science has made the older methods more effective and has also introduced new and highly reliable methods of birth control. The ability to control reproduction has led to a great many social changes for women and for men. People can now decide whether or not they want to have children. If they decide to have a family, the number of children and the time interval between babies can be planned.

Most methods of family planning, or birth control, involve prevention of fertilisation and hence of conception. Measures that prevent a woman from having a child are therefore usually called **contraception**.

Shutterstock.com/Rido

FIGURE 13.1
Contraception allows parents to plan their family

Abstinence

Abstinence is not having sexual intercourse at all. This is the only option that has no risk of either pregnancy or side effects.

Detection of ovulation

Some methods of birth control rely on a female's ability to determine the time of ovulation. She can then abstain from sexual intercourse on the days when fertilisation is most likely. This is known as **periodic abstinence** or the 'safe period' technique. There are a number of ways of determining the safe period.

Rhythm method

The **rhythm method** is based on the fact that an egg is available for fertilisation during a period of only three to five days in each menstrual cycle. If a female has a regular 28-day menstrual cycle, ovulation is likely to occur on about the 14th day. As the egg can survive for only two days unless it is fertilised, and sperm can survive in the female reproductive tract for four days at the most, sexual

The theory of rhythm: 28-day cycle

1 Menstruation begins	**2**	**3**	**4**	**5**	**6**	**7**
8	**9**	**10**	**11**	**12**	**13**	**14**
		Intercourse on these days leaves live sperm to fertilise the egg		Ripe egg may be released on any of these days		
15	**16**	**17**	**18**	**19**	**20**	**21**
Ripe egg may also be released on these days		Egg may still be present				
22	**23**	**24**	**25**	**26**	**27**	**28**
1 Menstruation begins again						

White squares – 'safe days', when conception is unlikely
Coloured squares – 'unsafe days', when pregnancy may occur

FIGURE 13.2 The 28-day cycle showing 'safe days'

intercourse should not occur between four days before and four days after ovulation, if pregnancy is to be avoided. An extra allowance should then be made in case the egg is not released exactly on day 14, but a day or so earlier or later. Figure 13.2 illustrates the safe and unsafe days for a 28-day cycle. Most women do not have cycles that are exactly the same each month, and the cycle is seldom exactly 28 days. For this reason, the rhythm method is now usually used in combination with the other methods described below.

Temperature method

The **temperature method** is a refinement of the rhythm method of predicting ovulation. A female can take her body temperature each morning to determine the time of ovulation more accurately. Ovulation is accompanied by a sharp drop in body temperature and then a rise, as shown in Figure 13.3. Using this method, a woman then knows she can safely have intercourse three days after the temperature rise has occurred.

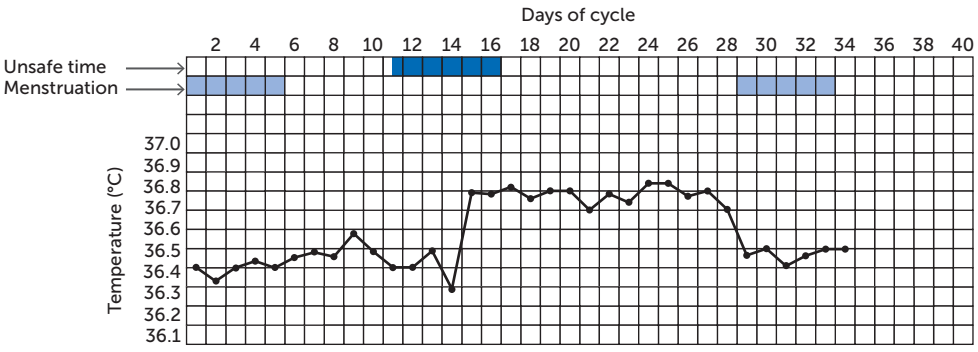


FIGURE 13.3 Body temperature and the ovarian cycle

Mucus method

The **mucus method**, developed by an Australian doctor, is another way of predicting the safe period more accurately. The probable time of ovulation is predicted by observing a change in the **mucus** of the cervix. Immediately after menstruation the tissues of the vaginal opening feel dry. As ovulation approaches, mucus can be detected. At first it is cloudy and sticky, but as the cervix secretes more mucus its nature changes: the mucus becomes clearer, feels slippery to the touch, and strands will stretch without breaking. On the day of ovulation the peak of clear mucus is reached, after which it becomes cloudy again. Sexual intercourse is 'safe' when there is no mucus, and more than three days after the last day of the clear mucus.

9780170449090

Symptothermal method

The **symptothermal method** uses the rhythm method and a combination of the temperature and cervical mucus methods to predict the fertile period of a female's cycle more accurately. A Fertility Monitor has been developed to measure daily changes in body temperature and cervical mucus.

None of these safe period methods, often referred to as fertility awareness methods, is particularly reliable and all rely on the woman keeping careful records. Another problem is that in a close relationship it is very difficult to have sex according to a calendar rather than according to desire. Close cooperation and support from the woman's partner is essential. However, if the couple are motivated and the woman is properly trained in the methods of detecting ovulation, very low pregnancy rates can be achieved.

Lactational amenorrhoea

Lactational amenorrhoea is the temporary infertility that follows the birth of a child. It occurs when a woman is not menstruating (amenorrheic) and is fully breastfeeding. Used as a means of contraception, it is known as the lactational amenorrhoea method (LAM). It relies on the fact that breastfeeding affects the production of hormones so that ovulation is suppressed. The chance of a pregnancy is therefore reduced. This method can be very effective, but only when a woman's menstrual periods have not returned, and the baby is being fully breastfed (i.e. is not being fed any food or milk supplements) and is less than six months of age.

Coitus interruptus

Coitus interruptus, or withdrawal, is the removal of the penis just before male orgasm so that ejaculation takes place outside the female vagina. This, the oldest method of contraception, is highly unreliable. It depends on the male being able to recognise the sensations that occur just before ejaculation and requires considerable self-control for withdrawal to occur in time. Even if the penis is withdrawn prior to ejaculation, some sperm may escape in the pre-ejaculatory fluids, so fertilisation could still occur.

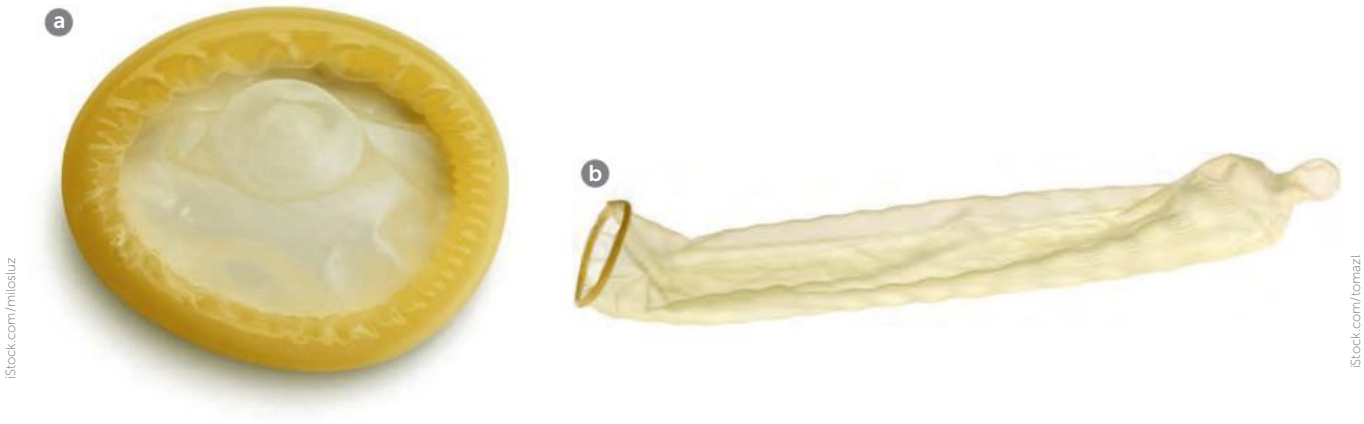
Mechanical barriers

A variety of mechanical barriers can be used to prevent the sperm from reaching the egg. This means that, when used correctly, they prevent fertilisation.

Condom

A **condom** is made from very thin latex rubber that is rolled on to the erect penis just before intercourse. There is some evidence that condoms made of animal membrane were in use over 2000 years ago. A condom is effective in preventing semen from entering the vagina, provided it does not tear or slip off after ejaculation. Condoms have an additional advantage in providing protection against sexually transmitted infections such as HIV/AIDS and syphilis. These will be discussed later in this chapter.

9780170449090



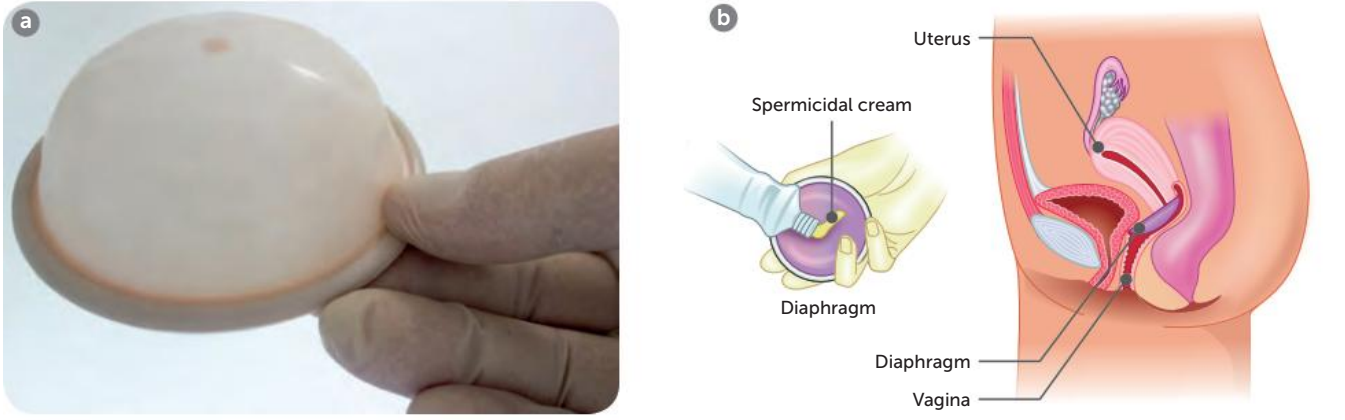
iStock.com/milosluz

iStock.com/tomazl

FIGURE 13.4 a Rolled-up condom; **b** Unrolled condom

Diaphragm

The **diaphragm** is a mechanical barrier used by the female. It is a thin rubber cap that fits across the top of the vagina. The correct size must be prescribed by a doctor. It must be inserted before intercourse, and is normally used with a spermicidal cream or jelly to increase effectiveness. It should be left in for at least six hours after intercourse.



iStock.com/Lalocracio

FIGURE 13.5 a Diaphragm; **b** Position of a diaphragm

Cervical cap

The **cervical cap** is similar to, but smaller than, the diaphragm. It fits directly over the cervix. Similar to the diaphragm, the cervical cap must be inserted prior to having intercourse and left in for at least six hours after ejaculation, and should be used with a spermicide.

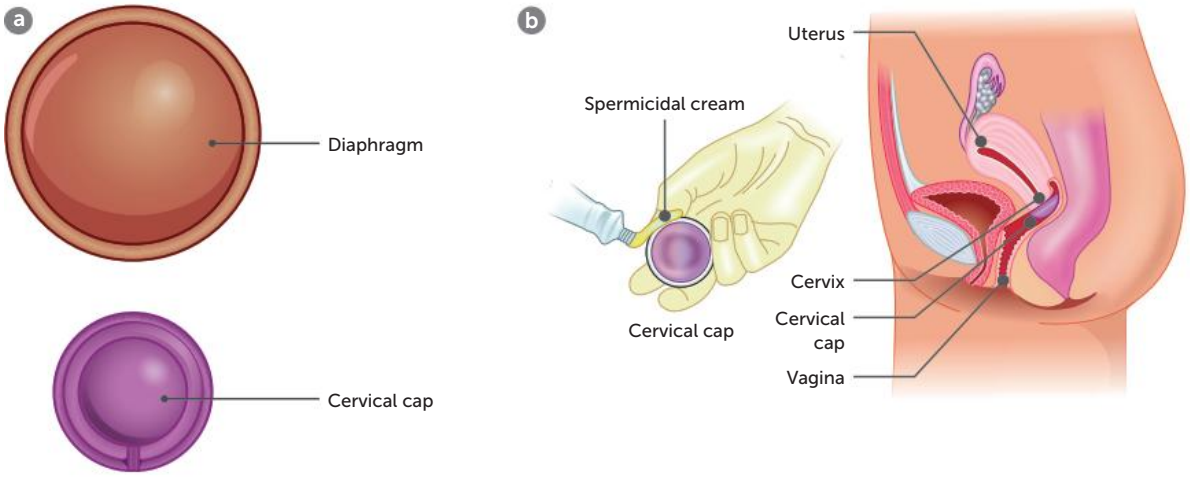


FIGURE 13.6 a Comparing a diaphragm and a cervical cap; **b** Position of a cervical cap

Female condom

The female condom (the **Femidom**) is a lubricated polyurethane sheath that lines the vagina. At each end of the sheath is a flexible ring; the one at the closed end fits over the cervix, and the other sits over the folds of skin that surround the entrance to the vagina. The female condom is an effective contraceptive device and gives protection against sexually transmitted infections.

Getty Images/Science & Society Picture Library

FIGURE 13.7
The Femidom, a condom for females

Spermicides

Spermicides work in two ways:

- They contain a substance that immobilises and destroys sperm.
- They react with moisture in the vagina to form bubbles of carbon dioxide gas, which present a physical barrier to the sperm.

They are available as creams, tablets, pessaries or aerosol foam.

Spermicides may be used with the condom, diaphragm and cervical cap. They are very unreliable when used alone, but they do add to the effectiveness of barrier devices.

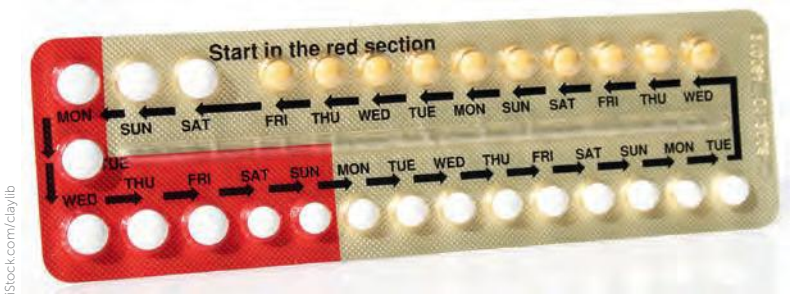
Hormonal contraception for women

One of the most effective methods of contraception is to prevent ovulation by changing the hormone levels in the female's body.

Contraceptive pills

The oral contraceptive pill, or 'the Pill', is the best-known form of hormonal contraception. Since their introduction in Australia in 1961, oral contraceptive pills have become widely used due to their reliability and convenience. Many different brands are available, but there are two main types.

FIGURE 13.8 Contraceptive pills



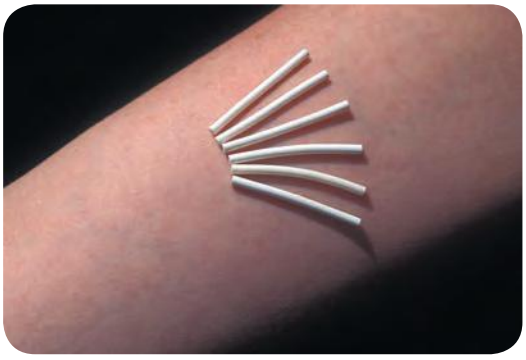
iStock.com/claylib

The first type, known as the **combined pill**, contains substances similar to the two female hormones, oestrogen and progesterone. When taken daily for the first 21 days of the menstrual cycle, the substitute hormones prevent the release of mature eggs from the ovary. The cervical mucus also becomes thick and sticky, making it difficult for sperm to travel upwards from the vagina. In addition, the 'hormones' alter the lining of the uterus so that it becomes less receptive to the implantation of an embryo. These three effects protect a female against an unwanted pregnancy, provided the pill is taken daily. If missed for more than two days, hormone levels drop and there will be no protection. Some brands use inactive pills for the seven days during which hormonal substitutes are not required. This means that a pill is still taken every day, making it less likely that one will be missed.

The second type of contraceptive pill, called the **mini pill**, contains only the progesterone substitute progestogen. This 'hormone' makes the cervical mucus thicker so that sperm cannot enter the uterus. It also changes the lining of the uterus, making it difficult for a fertilised egg to implant. The mini pill must be taken daily at the same time each day.

Depo-Provera and Depo-Ralovera are forms of the hormone that are injected into the muscle of the upper arm or buttock. The injection lasts for 12 weeks and works in a similar way to the mini pill.

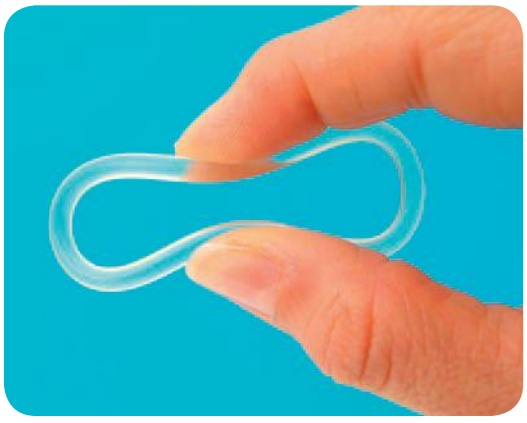
FIGURE 13.9 The hormone implant Implanon NXT


Alamy Stock Photo/Scott Camazine

Hormone implants

Another way of delivering progestogens is to use **Implanon NXT**. This is a soft plastic stick about 4 cm long that is inserted beneath the skin on the inner side of the upper arm. It slowly releases progestogen into the body and provides contraception for three years. The implant can be easily removed, and ovulation usually returns within three weeks.

FIGURE 13.10 The NuvaRing vaginal ring


Alamy Stock Photo/imageBROKER

Vaginal ring

Oestrogen and progesterone can also be delivered by a vaginal ring, marketed in Australia as **NuvaRing**. The soft plastic ring is placed in the vagina, where it releases low doses of the two hormones. It is left in the vagina for three weeks and taken out

for one week. The small amounts of hormones released prevent ovulation, cause the mucus in the cervix to thicken so that sperm cannot enter the uterus, and change the lining of the uterus so that implantation cannot occur. NuvaRing has the same hormones as the combined pill, and produces the same effects, but the woman does not have to remember to take a pill each day.

The oestrogen in the combined pill causes many of the side effects of hormonal contraception. Because the progestogen-only pills, injections or implants do not contain the oestrogen substitute, many of the side effects of the combined pill do not occur.

Despite the risk of side effects, the combined pill is currently one of the most reliable hormonal contraceptives available. Provided it is taken daily, it gives almost 100% protection. With continual research and development, the formulation of the combined pill has gradually changed, and the amount of hormone substitutes has been reduced considerably. This has resulted in a marked decrease in the incidence of side effects. The most serious side effect of the contraceptive pills now in use is the increased risk of developing a blood clot in a vein or an artery. Although small, the risk does increase with age. For women over the age of 35 the combined pill is quite safe as long as they do not smoke; if a woman wants to smoke, she should use some other form of contraception.

Hormonal contraception for men

Male hormonal contraceptives are being developed in a number of different forms. One that has been trialled in Australia involved an implant of the hormone testosterone being placed under the skin every four months. This was combined with an injection of progesterone every three months, and was found to be effective in suppressing sperm production. Ongoing research on 1000 men in China is designed to test the drug's effectiveness, evaluate its safety and monitor side effects. Other options currently under development include a gel or tablets that also contain testosterone and a synthetic progesterone.

Intrauterine devices

Intrauterine devices, or **IUDs** (sometimes called intrauterine contraceptive devices, IUCDs), are small devices made of plastic, and often containing copper, that are inserted into the uterus. Once an IUD is inserted into the uterus, it is not felt by the woman or her partner. All IUDs available in Australia have fine nylon threads attached to their lower end so that, when fitted, the threads extend through the cervix into the upper vagina. These threads allow a woman to check that the IUD is still in place and also allows for easy removal by a doctor.

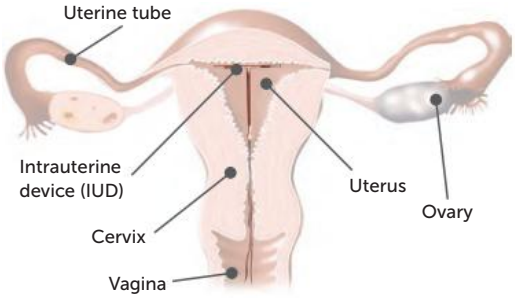


FIGURE 13.11 An IUD in the uterus

There are two main types of IUDs: hormonal and copper.

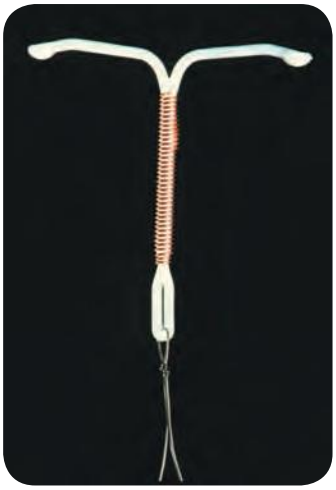
Hormonal IUD

A hormonal IUD is made up of a plastic frame with a core that slowly releases the progestogen hormone levonorgestrel. In Australia, there is currently only one type available, Mirena, which is sometimes referred to as an intrauterine system (IUS).

The hormonal IUD works by releasing its hormone at a steady rate that makes the lining of the uterus, the endometrium, thin and unsuitable for the implantation of a fertilised egg. The hormone also stimulates the cervix to produce thick mucus that prevents sperm from entering the uterus and swimming towards the egg. In some women, the hormone from the IUD stops ovulation altogether.

FIGURE 13.12 Hormonal IUD Mirena

9780170449090



Science Photo Library/Garry Watson

FIGURE 13.13
Copper IUD

Copper IUD

A copper IUD consists of a plastic frame with copper sleeves or copper wire around it. There are two types currently available in Australia.

Copper IUDs work mainly by inhibiting the movement of sperm and thus preventing them from moving through the uterus. In addition, they cause changes to the endometrium which, if an egg were to be fertilised, would stop the egg from attaching to it. Because of this, copper IUDs are sometimes used as an effective form of emergency contraception up to five days after unprotected sex.

The effectiveness of IUDs has improved greatly since the first IUD, known as the Lippes Loop, became available to women in 1964. They are now more than 99% effective and retain their effectiveness over a long period of time. Whereas Mirena provides protection for five years, copper IUDs are effective for up to 10 years. In addition, the devices can be removed at any time and fertility returns quickly.

Emergency contraception for women

Sometimes it is necessary to try to prevent pregnancy after sex instead of before. This could happen if a condom breaks, a pill is forgotten or rape occurs. The so-called **morning-after pill** was most commonly taken as two tablets of progestogen, but is now more commonly available as a single tablet. Since 2004, the emergency contraceptive pill (ECP) has been available over the counter at Australian pharmacies.

Emergency contraceptive pills have several different names, including Postinor, Levonelle or NorLevo-1. The sooner the pill is taken after unprotected sex, the more effective it will be; effectiveness is much reduced after 72 hours, although it may still prevent pregnancy if taken four or five days after sex.

The emergency contraceptive pill works by preventing or delaying ovulation, preventing sperm from reaching an egg, and preventing implantation of an embryo in the lining of the uterus.

There are few side effects. Nausea and vomiting may occur, but they are not common. Other possible, but rare, side effects are headache, stomach ache, breast tenderness, dizziness, or spot bleeding from the vagina.

In Australia, there has been concern from parents and others that, with the morning-after pill now available from pharmacies without a doctor's prescription, young girls can buy the pills over the counter from their local chemist. Parents may therefore be unaware that their children are using emergency contraception. This is one example of developments in human biological science that can cause tensions for individuals and society.

As mentioned previously, a copper IUD can be used effectively as emergency contraception in some women. It can be particularly appropriate for those who are considering using an IUD as a means of future contraception. IUDs interfere with the movement of sperm and change the lining of the uterus to prevent implantation of a fertilised egg. They can be 99% effective as emergency contraception if inserted within five days of unprotected sexual intercourse.

Sterilisation

Sterilisation is a permanent method of birth control for both men and women where the anatomy of the reproductive system is altered so that the sperm and egg are unable to meet. The choice of sterilisation as a birth control method for both men and women should be considered only when no further children are wanted. While it is able to be reversed in some people, it should be thought of as a permanent procedure.

9780170449090

Male sterilisation

The sterilisation operation in the male, called **vasectomy**, has traditionally involved the removal of a small piece of each vas deferens. The operation is relatively simple: a small cut is made on each side of the scrotum. A small segment is then removed from each vas deferens and the cut ends are tied or sealed with heat. The cuts in the scrotum are then closed. Most operations are done under local anaesthesia, but a general anaesthetic can be used.

Sterilisation does not result in any loss of sexual desire or pleasure.

A new, non-surgical vasectomy is being tested in animals with some success. This technique involves injecting a gel into the vas deferens that blocks the path of the sperm.

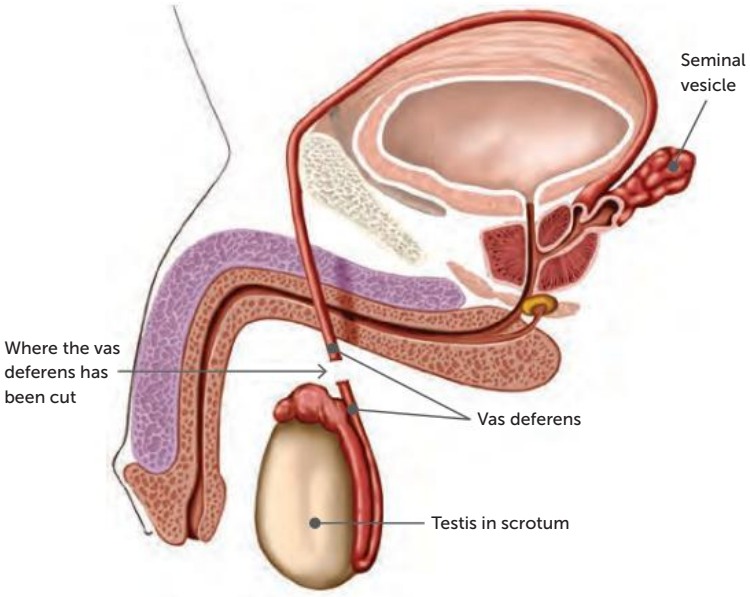


FIGURE 13.14 Vasectomy

Female sterilisation

Sterilisation in women is generally achieved by performing a **tubal ligation**, sometimes referred to as a tubal occlusion. This operation is a relatively simple procedure, requiring only a short stay in hospital. Under a general anaesthetic, a small incision is made in the abdomen and the uterine tubes are located. Each tube is then cut, a small piece is removed, and the ends are tied. Alternatively, an instrument called a laparoscope may be used. It is passed into the abdominal cavity through a small (1 cm) cut at the lower edge of the navel. Once it is inside the abdominal cavity, the doctor is able to locate the uterine tubes and fit metal clips to each, crushing that section of the uterine tube. After tubal ligation, sperm cannot reach the egg, and the egg cannot reach the uterus. A female has no decrease in sexual desire as a result of this operation.

The sterilisation techniques discussed here should not be confused with the removal of the sex organs. **Castration** is the removal of the testes, and **oophorectomy** is the removal of the ovaries. Both these operations affect the balance of the reproductive hormones and have profound effects on sexual drive and body characteristics. They are usually performed only when the organs are diseased. In women, the removal of the uterus, called a **hysterectomy**, also results in sterility.

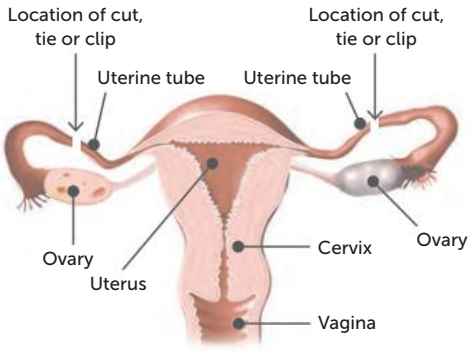


FIGURE 13.15 Tubal ligation

Key concept

Methods of contraception prevent the sperm and egg meeting. They vary in the reliability of their effectiveness as well as possible side effects.

Types of contraception
This website gives more details about different forms of contraception.

9780170449090

Contraception choices
This Australian website deals with contraceptive choices.

Choice of contraception methods

There are many factors to take into consideration when choosing a method of contraception. These include the reliability, side effects, convenience, availability, cost, permanence and personal preferences, including religious beliefs.

Table 13.1 summarises some of the advantages and disadvantages of the various methods.

TABLE 13.1 Advantages and disadvantages of various methods of birth control

METHOD OF BIRTH CONTROL	ADVANTAGES	DISADVANTAGES
Natural methods	No side effects; no costs; acceptable to certain religious groups	Poor reliability; these methods provide no protection against sexually transmitted infections (STIs)
• Periodic abstinence (safe period)		Time and effort required to determine ovulation; not very reliable; abstinence required at certain times
• Lactational amenorrhoea method		Relies on fully breastfeeding a child; effective only with no menstruation and within first six months of birth
• Withdrawal		Requires self-control; very unreliable
Spermicides	Relatively easy to use	Very unreliable on their own; need to be used in conjunction with another barrier such as a diaphragm or cervical cap; no protection against STIs
Intrauterine devices	Effective; long lasting; easily reversed; once in place can be forgotten; in some women, may be effective emergency contraception	Must be inserted by doctor; in some women cause pain and bleeding at menstruation; no protection against STIs
Mechanical barriers		
• Diaphragm and cervical cap	Does not affect the menstrual cycle; can be used during menstruation; can be inserted ahead of time so that spontaneity of intercourse is not affected	Difficult or unpleasant to insert; correct size must be prescribed by doctor; spermicide must be used to improve reliability
• Condom	Easy to buy; relatively cheap; good protection against HIV and other STIs	May affect spontaneity; partners need to be motivated and cooperative
• Femidom	May be put in place long before intercourse; stronger than male condoms; good protection against HIV and STIs	Placement needs practice; more expensive than male condoms
Hormonal contraception for women (None of the hormonal methods provide protection against STIs)		
• Combined pill	Very reliable; regular periods; reduced incidence of ovarian and uterine cancer; unrelated to sexual activity	Regular doctor's prescription required; pill must be taken daily; possible side effects
• Mini pill	Reliable if taken carefully; suitable for women who cannot take oestrogen	Must be taken at same time every day
• Implanon NXT	Lasts three years; relatively cheap; nearly 100% effective	May cause menstrual irregularities; possible side effects
• Depo-Provera and Depo-Ralovera	Very effective; convenient; periods cease	Injection cannot be reversed; delay in return to fertility when injections cease; possible side effects
• NuvaRing	Daily pill not required; very reliable	Regular placement and removal required
Sterilisation – tubal ligation and vasectomy	Permanent; nearly 100% effective	Cannot be easily reversed; require a surgical procedure; specialist referral necessary for female sterilisation; no protection against STIs
Morning-after pill	May be useful when other methods have failed or have not been used; fairly effective; available over the counter	Emergency use only; needs to be started within 72 hours of sexual intercourse to be effective; no protection against STIs

9780170449090

Reliability

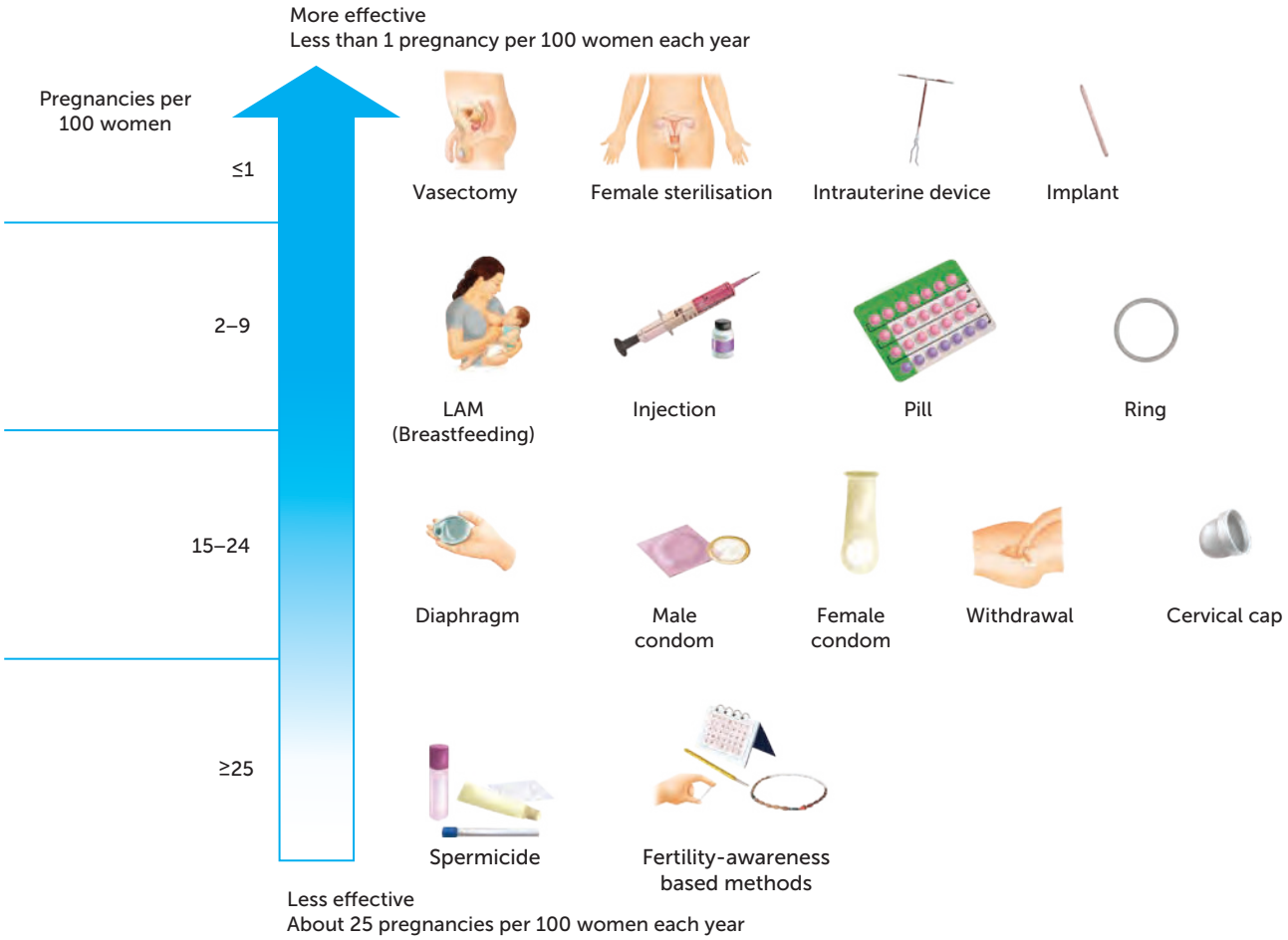


FIGURE 13.16 Relative effectiveness of birth control procedures

Ethical issues

There are several ethical issues associated with birth control. For example, the Catholic Church, and some other Christian faiths, hold the view that the only acceptable method of family planning is abstinence from intercourse at times when fertilisation is most likely to occur.

Some people believe that methods that allow fertilisation but prevent implantation, such as IUDs and morning-after pills, are morally wrong. The arguments against these methods centre on the question of when an embryo becomes a human being: is it at the moment of fertilisation, the time of implantation, or some later stage of embryonic development?

Questions relating to our beliefs and values cannot be answered by science. Each of us must consider the implications carefully and make our own decisions about such matters.

Key concept

The choice of contraception is a personal one and is based on factors such as reliability, availability, cost and side effects, as well as ethical and moral considerations.

Activity 13.1
Researching developments in contraception

9780170449090

Questions 13.1

RECALL KNOWLEDGE

1. State the purpose of contraception.
2. Name the only type of contraception that is 100% effective.
3. Describe the methods of detecting ovulation.
4. Describe the changes in body temperature that occur at the time of ovulation.
5. a Explain why coitus interruptus is also called the withdrawal method.
   b Explain why it is an unreliable form of contraception.
6. List the contraception methods of mechanical barriers.
7. Compare and contrast the diaphragm and cervical cap as methods of contraception.
8. List the hormones that are used in contraception for males and females. For each hormone, state how it achieves its function.
9. What does the abbreviation 'IUD' stand for?
10. Describe how a vasectomy and tubal ligation prevent pregnancy.
11. List the factors that should be considered when selecting a method of contraception.

APPLY KNOWLEDGE

12. Explain why it is important not to have sexual intercourse for a few days prior to ovulation in order to avoid pregnancy.
13. Lactational amenorrhoea is the temporary infertility that occurs following the birth of a child if the mother is breastfeeding.
    a Suggest why lactational amenorrhoea is an advantage for the mother and child.
    b Suggest why mothers who are not breastfeeding should not rely on lactational amenorrhoea.
14. State three reasons why pregnancies can still occur when condoms are used. For each reason, explain how it can lead to pregnancy.
15. The contraceptive pill uses hormones to prevent pregnancy.
    a Suggest why hormonal methods of contraception are one of the most effective methods of contraception for females.
    b Discuss situations where this effectiveness is reduced.
16. Discuss the advantages of a hormone implant over hormone pills.
17. Suggest why some people do not agree with the use of the morning-after pill as a form of contraception.

13.2 SEXUALLY TRANSMITTED INFECTIONS

Sexually transmitted infections (STIs), formerly referred to as **sexually transmitted diseases (STDs)**, are infections that are transmitted by close body contact, usually with the genital organs. They are caused by viruses, bacteria, fungi or parasites that are passed from an infected person to a partner during sexual activity.

Young people are most at risk of contracting an STI. In Australia, three-quarters of known cases of STIs occur in people aged 15 to 29 years. Unlike many other communicable diseases, there is no vaccine readily available for the majority of STIs. Furthermore, for many STIs, one attack does not make a person immune or any less likely to contract the disease if exposed to infection again.

9780170449090

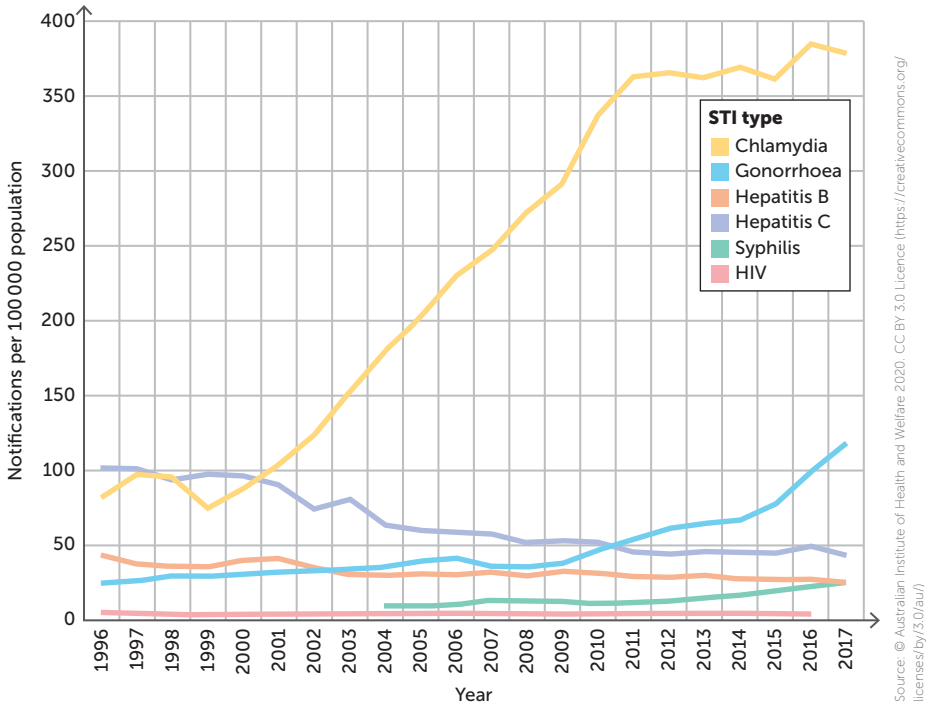
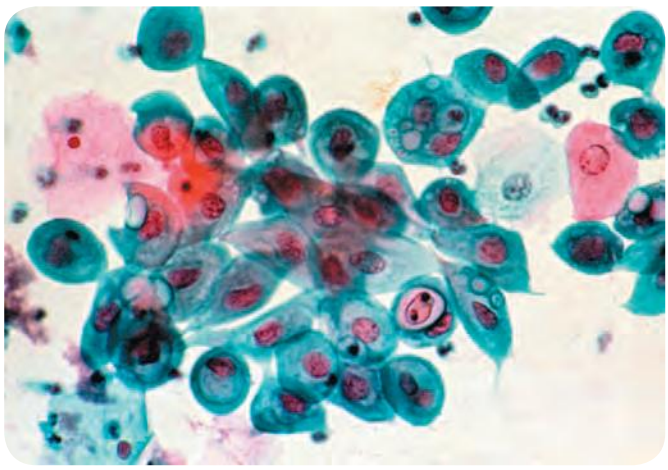


FIGURE 13.17 Notification rates of sexually transmissible infections and blood-borne viruses in Australia, 1996–2017

Chlamydia

The most common STI, **chlamydia**, is caused by a highly specialised bacterium, *Chlamydia trachomatis*. This bacterium is only able to reproduce inside a living human cell, making it difficult to isolate. Most people show no symptoms and are unaware that they are infected with chlamydia. It is therefore sometimes called the 'silent infection'. When an infection is diagnosed, it can be treated with antibiotics.

The incidence of chlamydial infection in Australia has risen dramatically, as shown in Figure 13.17. Although there were 378.6 reported cases per 100 000 people in 2017, many cases go unreported and some health experts suggest that there are actually many more cases.



Science Photo Library

FIGURE 13.18 Photograph of a cervical smear taken with a light microscope. Spherical *Chlamydia trachomatis* cells (dark pink) can be seen inside the epithelial cells of the vagina (light pink and light blue). The bacterium reproduces inside the host cell and the new bacteria are released by rupturing the host. This can be seen occurring at the bottom left corner

Chlamydia infection

Chlamydia trachomatis is transmitted by vaginal or anal sex with an infected person. Both men and women may be infected, and it occurs mostly in people aged under 25 years.

Men may develop an infection of the urethra known as **non-specific urethritis (NSU)**, which has the symptoms of a yellow, mucus-like discharge from the penis and a burning sensation when passing urine. About half the known cases are thought to be caused by *Chlamydia trachomatis*. Other organisms that can cause NSU are *Escherichia coli* (a bacterium), *Candida albicans* (a fungus) and *Herpes simplex* (a virus).

Incidence of sexually transmissible infections
This website has statistical data about STIs in Australia.

Healthy behaviours
This website looks at healthy behaviours for children, including sexual behaviours.

STIs – Get the Facts
This Western Australian website gives more details about STIs.

Keeping Safe – Get the Facts
This Western Australian website gives more details about keeping safe.

9780170449090

If the chlamydia infection in males is not treated, the bacterium can spread to the epididymis, where it causes inflammation. This inflammation, called **epididymitis**, causes pain and swelling of the epididymis. In Australia, chlamydia is the most common cause of epididymitis in men under the age of 35. It can lead to infertility if both testes are infected. However, epididymitis in both testes is very rare.

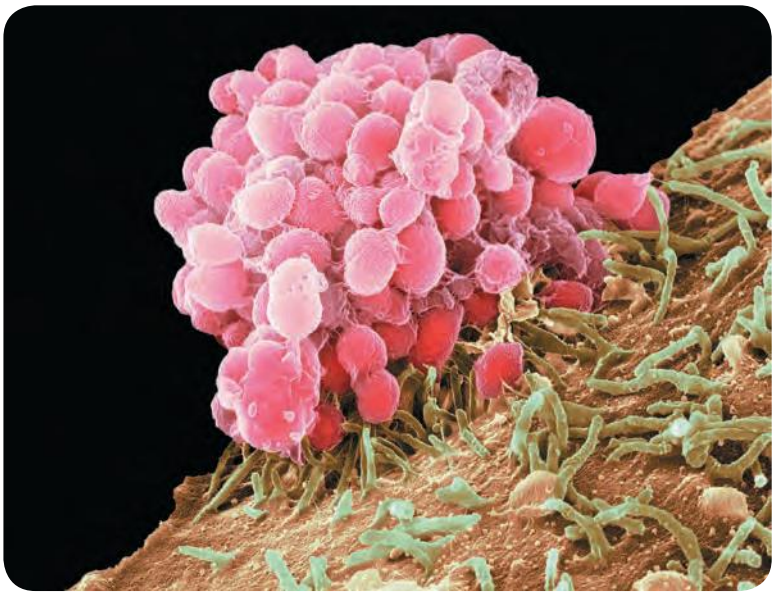
Most infected women have no symptoms and therefore have no idea that they have chlamydia. This makes the disease very dangerous, because if untreated it can lead to infertility, eye infection and arthritis. Some women show symptoms of **pelvic inflammatory disease (PID)**, which is inflammation of the organs in the pelvic region such as the uterus and uterine tubes. Continual inflammation of the uterine tubes may lead to blockage by scar tissue, and thus to infertility. Implantation of an embryo outside the uterus – an **ectopic pregnancy** – may also occur. If the infected woman is pregnant, there is a 70% chance that the disease will be passed to the foetus during birth. The baby may then suffer from conjunctivitis, nose and throat infections, or pneumonia. There is also some evidence that chlamydia may cause a significant increase in the risk of having a premature birth or a stillborn child.

Diagnosis and treatment of chlamydia

Correct diagnosis and treatment of chlamydia is vital. If a chlamydial infection is suspected, a urine test can be done in males and in females. A more accurate diagnosis can be made with a swab from the vagina, cervix, anus or penis, which is then analysed in a laboratory. If infection by *Chlamydia trachomatis* is confirmed, the usual treatment is a course of antibiotics, although even with prolonged treatment the bacterium may never be completely eliminated from the body.

Research into a vaccine against chlamydia is being carried out in Australia and elsewhere. Researchers at the Queensland University of Technology have developed a vaccine that prevents infertility from chlamydia in mice. It is an innovative approach in that it does not aim to vaccinate against contracting the disease, but instead builds the body's tolerance to the bacterium. In this way, the vaccine prevents the presentation of negative symptoms such as infertility. However, a vaccine for use in humans is still at least a decade away.

It is interesting to note that the same bacterium that causes the sexually transmitted chlamydial infection also causes the eye disease trachoma. Trachoma is endemic in 51 countries and is the main cause of infectious blindness. It has largely been eliminated from developed countries due to improved standards of sanitation and hygiene, but it still persists in developing countries.



Science Photo Library

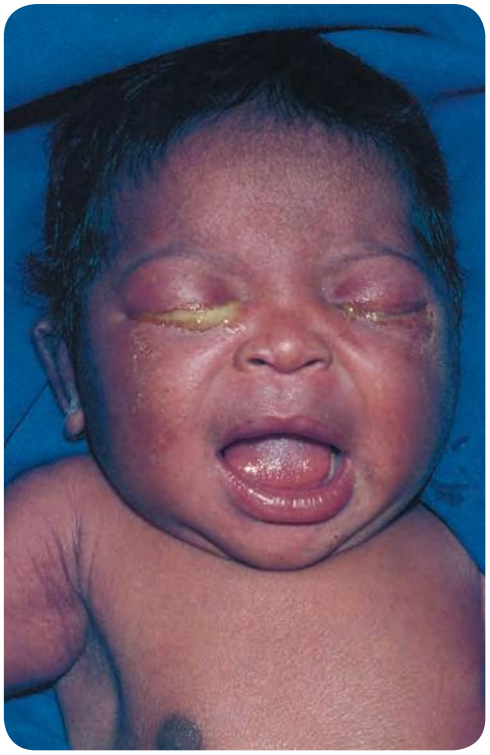
FIGURE 13.19 Scanning electron micrograph of *Neisseria gonorrhoeae* bacteria (pink) infecting a cell (brown)

Gonorrhoea

Gonorrhoea is an infectious disease that mainly affects the mucous membranes of the excretory and reproductive systems, the rectum, and occasionally the eyes and throat. The disease is caused by a bacterium, *Neisseria gonorrhoeae*, that is transmitted during sexual intercourse. The disease affects both males and females, and symptoms appear about 2–10 days after infection, although in females these may not be recognised. The period between infection and the appearance of symptoms is called the **incubation period** of a disease.

In males, the bacterium enters the urethra during intercourse with an infected partner. After the incubation period, inflammation in the urethra results in a burning feeling in the penis and extreme pain when passing urine. Later, there is a yellow discharge of pus from the penis. If untreated, the urethra may become permanently constricted, resulting in difficulty in urinating. If untreated, the infection can spread to other organs of the body, such as to the testes, causing eventual sterility; to the joints, causing a type of arthritis; or to the heart or eyes.

In females, the disease is sometimes considered more serious, as there may be no early symptoms, or the symptoms may go unrecognised in more than 70% of infected women. Bacteria enter the vagina during intercourse with an infected male. In most cases, the urethra or cervix are infected. There is usually no pain, so any pus produced is usually taken to be normal vaginal discharge. A woman may have the disease for months before pain causes her to seek medical treatment. Untreated, the infection spreads to the oviducts and to the abdominal membranes. Oviduct infection may cause permanent blockage and thus infertility. Because of the seriousness of an untreated infection and the fact that symptoms frequently do not appear, women who have sexual intercourse with a number of partners are advised to have smears taken on a regular basis to check for possible infection.



Science Photo Library/Dr MA Ansary

FIGURE 13.20 A baby's eyes ooze pus due to a gonorrhoeal infection

Discharge from the affected mucous membranes is the source of infection, and the bacteria are transmitted by direct contact, usually sexual. Therefore, oral sex with an infected person can result in infection of the throat. As gonorrhoeal infection in women occurs in the cervix and the vagina, children born to women with the disease may be infected during birth. The bacteria may enter through the baby's eyes, causing an acute eye infection that can lead to blindness.

Antibiotics are used in the treatment of gonorrhoea and normally result in an easy and effective cure, provided treatment is begun early enough. However, strains of gonococcus that are resistant to antibiotics have evolved and in some cases are becoming increasingly difficult to cure. These resistant strains are present in Australia. In early 2018, two cases of multi-drug resistant gonorrhoea were identified. These infections were resistant to all antibiotics.

Syphilis

Syphilis, also known as 'the pox', is caused by a thin, flexible, spiral-shaped bacterium, *Treponema pallidum*. The disease is normally contracted by direct sexual contact and affects men and women in the same way. The bacterium can only survive for a brief time outside human tissues; therefore, infections by indirect contact are extremely rare.

Stages of infection

The course of syphilis follows four stages if it is untreated.

1. The *primary stage* of syphilis begins when the syphilis bacterium enters through any small break in the skin. Such infection takes place during sexual activity with an infected person. The bacteria multiply and spread through the whole body during an incubation period ranging from 10 days to 10 weeks. The first symptom is one or more small sores, known as **chancres** (pronounced 'shankers'). These usually appear on the sex organs but may occur elsewhere on the skin such as lip, finger or eyelid. Chancres range from being so small that they are not noticed, up to 1 cm in diameter. The chancre heals in three to eight weeks, even without treatment, a situation that gives the infected person a false sense of security.

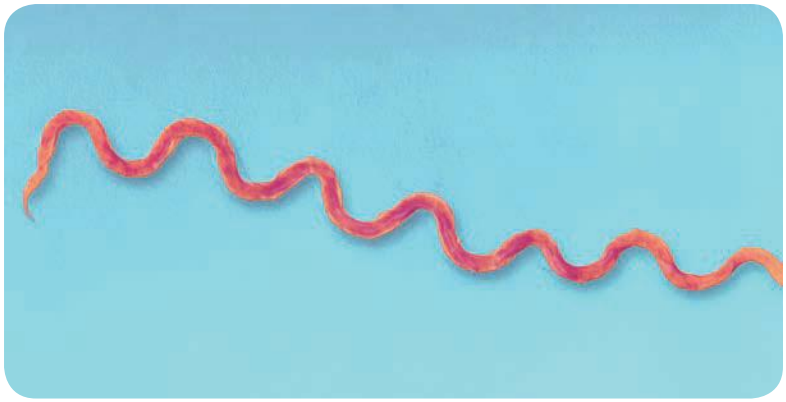


FIGURE 13.21 Coloured transmission electron micrograph of the spiral-shaped bacterium that causes syphilis

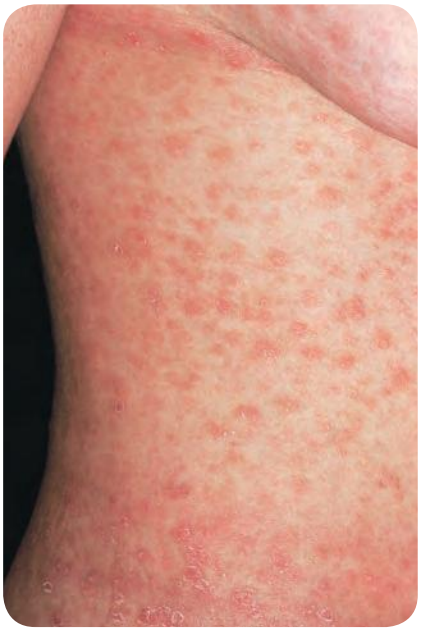


FIGURE 13.22 Skin rash during the secondary stage of an infection of syphilis

2 The *secondary stage* usually follows a few weeks after the primary stage, but may be delayed for up to 12 months. There is a large range of possible symptoms during this stage, including skin rashes, sore or ulcerated mouth or throat, mild fevers, and disorders of the bones or eyes. In some cases the symptoms are quite mild and may not be taken seriously. The skin rash due to syphilis persists for several weeks; any rash that appears and fades over a few days is not due to the disease. The patient is highly infectious during the secondary stage of the disease. This stage lasts about two years and all symptoms eventually disappear even without medical treatment.

3 A **latent stage**, or *hidden stage*, then follows. This begins when symptoms from the primary and secondary stages go away. The latent stage has no noticeable symptoms, but the body is still infected. It may last for many years, and in some cases for the rest of the person's life. During this latent period, the infection cannot be passed on to others. However, sometimes the symptoms of the secondary stage return. If this happens, the infection can then be passed to others while the symptoms persist.

4 Even without treatment, only a minority of people infected with syphilis develop the complications associated with the *tertiary stage*, or *late stage*. However, when symptoms do appear again the results may be devastating: syphilitic heart disease, insanity, blindness, weakening of the blood vessels, physical incapacity, and many other serious afflictions. This can occur anywhere between 5 and 40 years after the initial infection.

A special problem for pregnant women with syphilis is that bacteria can cross the placenta and infect the developing foetus. Infection of the woman is not necessarily followed by foetal infection, but once the bacteria enter the foetal circulation there is nothing to stop their multiplication. The foetus can go through all the stages of syphilis before birth, and may suffer permanent damage to the heart, nervous system, joints or other organs. Blood tests for syphilis are usually carried out during the pregnancy, and antibiotics can be used to treat both the infected woman and the foetus.

Syphilis in Europe
This website looks at the evidence regarding the introduction of syphilis into Europe.

Treatment

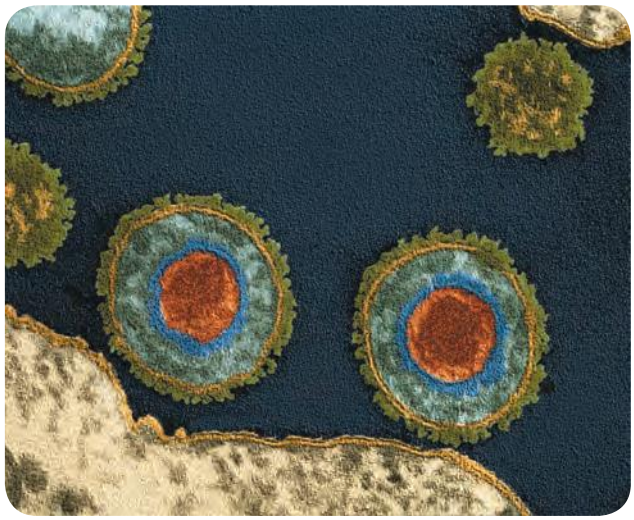
Antibiotics are the usual treatment for all stages of syphilis, and during the primary stage, cure is relatively easy. Surgery becomes necessary in the later stages.

Genital herpes

Genital herpes is a common STI caused by the **Herpes simplex** virus. There are two forms of the virus.

- Herpes simplex type 1 (HSV1) usually produces 'cold sores' on the lips but can also affect the genitals.
- Herpes simplex type 2 (HSV2) produces blisters on the genital organs.

Both types of the virus are transmitted by skin-to-skin contact and can therefore be passed on during genital, oral or anal sex.



Science Photo Library/Eye of Science

FIGURE 13.23
Transmission electron micrograph of the herpes simplex virus. The orange core is the viral DNA

The first episode of genital herpes is usually the most severe and can be very painful and distressing. Blisters develop in areas such as the penis of males, and the labia and vagina of females. There may also be accompanying flu-like symptoms or a rash. The blisters break, forming ulcers that then develop scabs. Healing occurs over a period of one or two weeks. Although healing has occurred, some of the virus passes into the nervous system, where it remains for life. The virus can then reinfect the skin or mucous membranes of the genital organs at any time, and the blisters can recur for the rest of the person's life. Recurrent episodes are not normally as painful and are of shorter duration than the first attack. It is important to note that the virus can still be passed on to others even when there are no symptoms.

The Herpes simplex virus can be transmitted from an infected mother to a baby during birth. In children born to infected mothers, serious malformations and life-threatening diseases may occur, although these complications are not common.

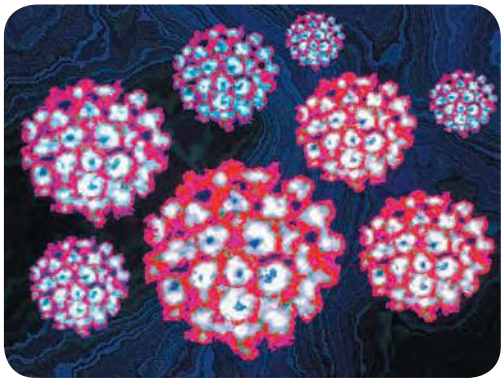
There is no way of removing the herpes virus from the body once infected. The treatment for genital herpes includes medication to reduce the pain, saline dressings to clean up the blisters, and sexual abstinence for the duration of the eruption. Antiviral drugs specific to herpes can be prescribed. They do not cure the disease but act to reduce the severity and length of an attack.

Genital herpes
This website has more information about genital herpes.

Herpes and pregnancy
This website discusses the risk of herpes during pregnancy.

Genital warts

Genital warts are usually found on the genital area – the vagina, labia, cervix or penis – but may occasionally be found around the anus or in the throat. They may be flat, raised or cauliflower-like growths. The warts are caused by a virus, the **human papillomavirus (HPV)**. More than 100 types of HPV have been identified and only a few of these cause genital warts. Some other types of HPV cause ordinary skin warts that can also occur on the genitals, but in such cases sexual transmission is not likely to be the cause.



Science Photo Library/Dr. Linda Stannard, UCT

FIGURE 13.24
The human papillomavirus

Genital warts are passed on by a sexual partner infected by the virus. As the warts may be inside the vagina or penis where they cannot be seen, there is a risk of being infected by a person who is unaware of the infection. A newborn child can become infected during passage through the birth canal.

Some types of HPV can cause cancer of the cervix, but those that cause genital warts *do not* cause cervical cancer.

Human immunodeficiency virus

Activity 13.2
Investigating the origin of HIV

Most people have heard of HIV and AIDS. The abbreviation '**HIV**' stands for **human immunodeficiency virus** and is known to cause **AIDS**, or **acquired immune deficiency syndrome**. HIV/AIDS is now **pandemic**, meaning that it has spread to all parts of the world. Infection with the human immunodeficiency virus weakens the body's immune system, and so the infected person is susceptible to infection by other micro-organisms and to some forms of cancer. Almost all cases of HIV infection eventually result in AIDS. *Acquired* means that the condition is not inherited, and *syndrome* refers to a set of symptoms or illnesses that occur as a result of one cause – in this case, an infection by HIV.

HIV and AIDS
This Australian website has more information about HIV and AIDS.

More information about HIV
This website contains information about all aspects of HIV and AIDS.

The origin of HIV

HIV infection

HIV is a **retrovirus**, containing an RNA core rather than a core of DNA. It is similar to other viruses in that it is unable to reproduce by itself. HIV infects white blood cells known as helper T-lymphocytes. Inside the host cells the virus uses an enzyme, reverse transcriptase, to convert its RNA into DNA. The DNA then integrates itself into the lymphocyte's DNA and produces millions of copies of itself. These new viruses are released into the blood to infect more T-lymphocytes. Some infected helper T-lymphocytes go into a resting, or latent, state. These cells act as a reservoir of HIV, as they are able to become active at any stage.

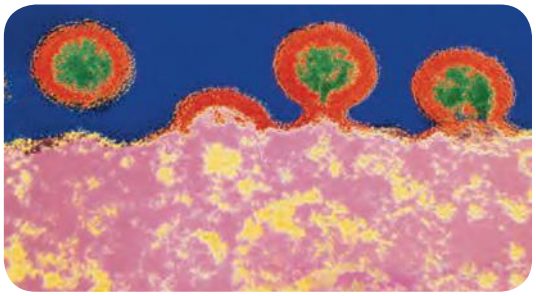
HIV infections go through a number of stages:

1. *Acute infection.* When a person is first infected with HIV, the virus replicates rapidly and the level of HIV in the blood is high. During this time, an infected person may suffer flu-like symptoms. The immune response overcomes the infection at this stage and the patient recovers. This stage lasts two to three weeks.
2. *Chronic infection*. The second stage is asymptomatic, where the person appears fit and well. The virus continues to multiply, but at a low rate. This stage may last 10 years or longer.
3. *AIDS*. A person develops AIDS when the immune system has been damaged to the degree that it is no longer able to resist other infections. At this stage, the person has a high viral load and a low level of helper T-lymphocytes.

The spread of HIV

HIV is transferred when body fluid from one person enters the bloodstream or comes in contact with the mucous membranes of another. The virus does not survive for long outside the human body and is not transmitted through air or water. The body fluids that are able to carry enough HIV to be infectious are blood (including menstrual blood), seminal fluid (including pre-ejaculatory fluid), vaginal and cervical fluids, and breast milk. Unless blood is present, the virus is not found in sufficient quantities to be infectious in urine, faeces, vomit, tears, sweat or saliva. Therefore, normal social contacts such as hugging, kissing and handshaking will not spread HIV. There is also no risk of getting the virus from activities such as playing sport, swimming in a pool with others, or from such things as coughing, sneezing, or sharing cutlery or crockery. HIV may be spread:

- by unprotected sexual intercourse (vaginal, anal or possibly oral) with an infected person
- by the sharing of needles and syringes with an infected person



Science Photo Library/NIBSC

FIGURE 13.25 Human immunodeficiency virus (HIV) may be transmitted by sexual contact. This coloured transmission electron micrograph shows HIV budding from an infected T-cell. The T-cell (pink) is at the bottom and four viruses can be seen in different stages of budding. Each virus has a protein coat (red) surrounding a core of RNA (green)

- from an infected mother to her child during pregnancy, childbirth or through breastfeeding
- by blood transfusions in countries where blood is not carefully tested
- by implements that pierce the skin if they have not been sterilised – for example, equipment used for ear and body piercing, tattooing, and for medical or dental procedures.

The following preventive measures may reduce the risk of becoming infected with HIV:

- choosing not to have sexual intercourse or to inject drugs
- having protected sex, unless both people are definitely free of HIV
- never sharing things that are likely to have human blood on them, such as needles or syringes.
- washing and covering any open cuts or sores on the skin and not allowing them to come into contact with human blood.

Diagnosis of HIV infection

If a person thinks they may have been infected with HIV, a treatment called post-exposure prophylaxis (PEP) can be given. The treatment, consisting of a course of drugs that must be taken for a month, must be started within three days of the possible exposure to HIV. The drugs prevent the replication of the virus so that it does not become established in the body. It is not 100% effective.

The body responds to infection with HIV by producing chemical substances called antibodies. Infection with HIV can therefore be diagnosed by tests that detect the presence of an HIV antibody in the blood. Depending on the individual, it takes from two to 12 weeks for the antibodies to build up to the point where they can be detected. If a person believes they have put themselves at risk of acquiring the infection, they should have a test at least two weeks after the possible exposure. Even then, if the test result is negative, a second test 10–12 weeks later is recommended, to be sure that no antibodies have developed.

In Australia, other tests for HIV are also available that measure viral load. 'Viral load' is the term used to describe the amount of HIV in the blood and gives an indication of the activity of the virus. The activity of the virus can be determined from the rate at which the virus replicates, and gives a guide to the likelihood of damage to the immune system. The results of viral load tests are given in terms of the number of viral copies of HIV per millilitre of blood.

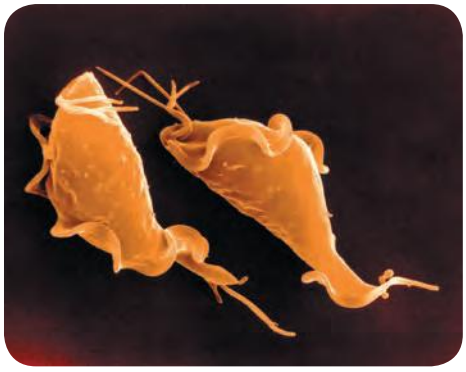
Treatment

Currently, there is no cure for HIV infection or a vaccine to prevent infection; however, there is a lot of research being conducted into these possibilities. One challenge that researchers are facing is the HIV reservoirs. Unless both the active and latent cells are able to be destroyed, an infected person cannot be cured.

There are, however, a number of antiretroviral drugs that inhibit the reproductive cycle of the virus. These drugs are divided into different classes, depending on which stage of the viral life cycle they affect. Patients are usually given a combination of many drug classes – a so-called cocktail of drugs. The content of a patient's multi-drug therapy depends on how high the viral load is, and whether they have had previous treatment with some of the drugs to which the virus has developed resistance.

These combination therapies have resulted in slowing the replication of HIV and suppressing the progression of the disease for some people. In certain cases, they reduce the viral load to levels that cannot be detected. However, the virus still remains in the body in such tissues as the spleen and lymph nodes. Therefore, therapy must be continued indefinitely.

HIV treatment and cure
This website includes a video that looks at current research into developing a cure or treatment for HIV.



Science Source/David M Phillips

FIGURE 13.26 *Trichomonas vaginalis*, the protozoan that causes trichomoniasis

Trichomoniasis

Trichomoniasis is an infection caused by a protozoan, *Trichomonas vaginalis*. It causes inflammation of the mucous membranes of the vagina in women and of the urethra in men. In women it causes great discomfort,

with symptoms including vaginal discharge and severe vaginal itch. Men often have the infection without symptoms; however, even without symptoms a man can transmit the infection to his female partner. The disease can be cured quickly and easily with antibiotics. Both partners should be treated at the same time even if one has no symptoms.

Trichomoniasis is spread by vaginal intercourse, so use of a condom will prevent infection.

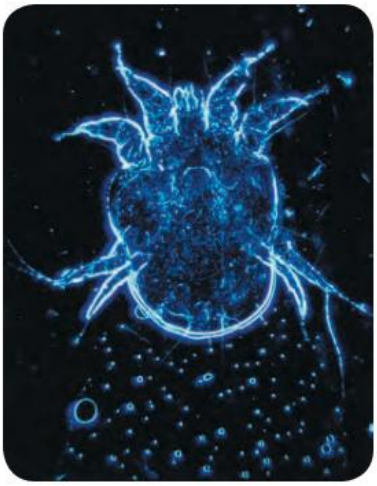
Pubic lice and scabies

Both pubic lice and scabies cause intense itching in the genital area. **Pubic lice** are insects called *Phthirus pubis*, which are broader than they are long. They are usually confined to the pubic and anal areas of the body, but in very hairy people may be found on the chest and in the armpits. The intense itching is thought to be due to an allergic reaction to the lice or to their faeces.

Scabies is caused by a mite, *Sarcoptes scabei*. Mites are members of the class Arachnida, which includes spiders and scorpions. They appear to prefer the genital region, wrists and finger webs, and these infected areas become extremely itchy, especially at night, often making sleep impossible. The itch may be due to sensitivity to the mite or to its faeces.

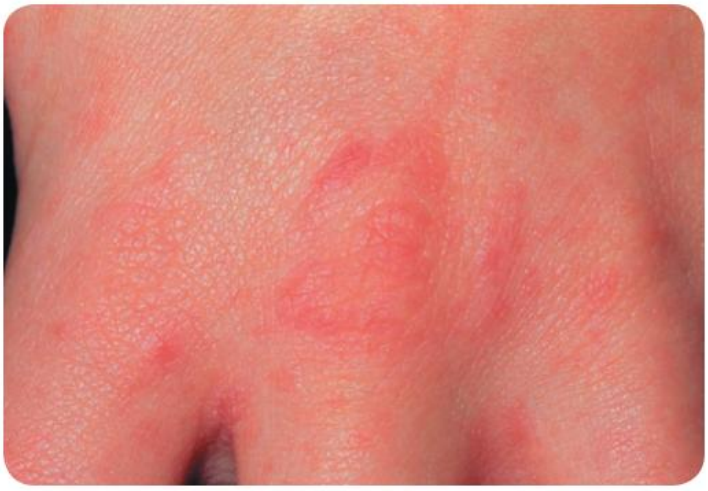
Science Photo Library/Eye of Science

FIGURE 13.27 Scanning electron micrograph of two pubic lice attached to human hairs. Note the large claw on each leg enabling them to cling to the hair



Science Photo Library/JC Revy, ISM

FIGURE 13.28 Light micrograph of *Sarcoptes scabei*



Science Photo Library/Dr P Marazzi

FIGURE 13.29 Rash on the skin of the hand caused by the scabies mite

Both pubic lice (often referred to as 'crabs') and scabies ('the itch') can be transmitted by sexual contact. However, sexual contact is not necessary if the partners are sharing the same bed: transmission frequently occurs merely through prolonged, warm, close contact.

Pubic lice and scabies are both treated using lotions that are applied to the skin. The lotions kill the insects or mites and also their eggs.

WS

13.1 Sexually transmitted infections

Key concept

Sexually transmitted infections, including chlamydia, gonorrhoea, syphilis, genital herpes, genital warts, HIV, trichomoniasis, pubic lice and pubic scabies are transferred from one partner to another during sexual intercourse.

Control of sexually transmitted infections

The identification, treatment and tracing of STIs helps to control their spread.

Preventing STIs

Abstinence is the only method of completely preventing the transfer of sexual transmitted infections. However, practising safe sex can reduce the chances significantly.

Safe sex involves taking precautions to make sure that your partner's semen, vaginal fluids or blood do not enter your body and that your fluids do not enter your partner's body.

For vaginal or anal intercourse, safe sex involves the use of a condom. This prevents exchange of body fluids and has the added advantage that it prevents pregnancy. For oral sex, condoms or dental dams can be used. A dental dam is a square of very thin latex that can be used as a barrier during oral sex.

Additionally, any parts of the body that may be infectious, such as warts or herpes sores, should be covered and contact with those parts avoided.

Safe sex and condoms
This website has information on safe sex and using condoms.

STI fact sheets
This website has links to detailed fact sheets about the different STIs.

Treatment and contact tracing of STIs

There are special clinics for the treatment of STIs in most of the major cities in Australia, and free treatment is also available in all public hospitals. Treatment is strictly confidential, and the names of patients are never revealed. In addition to treatment, an important part of the work of STI clinics is **contact tracing**. By tracing the sexual contacts of patients, the clinics aim to locate and treat people who unknowingly have these diseases. In this way, the spread of the disease can be limited.

Another important role of STI clinics is education. If people know the dangers, and are aware of the symptoms, they are more likely to seek treatment early, when the disease is more easily cured and before they pass it on to others.

Activity 13.3
Understanding the social consequences of vaccines for STIs

Key concept

Safe sex prevents new STI infections by stopping the transfer of infected body fluids from one partner to the other. Control of the spread of STIs relies on the identification, treatment and tracing of infections.

Questions 13.2

RECALL KNOWLEDGE

1. List six sexually transmitted infections.
2. Which STI has had the highest incidence in Australia in the past two decades?
3. List the symptoms of chlamydia in males.
4. Describe the symptoms of genital herpes.
5. Name the cause of genital warts.
6. Describe the stages of HIV infection.
7. Explain why the use of condoms is effective in preventing the spread of HIV.
8. Pubic lice and scabies both cause itching. Explain why they cause this symptom.
9. Explain why gonorrhoea causes:
   a. pain during urination in males
   b. infertility in females.
10. Use a flow chart to summarise the stages of syphilis infection.

APPLY KNOWLEDGE

11. Suggest why young people are more at risk of contracting an STI than older people.
12. Explain why it is difficult to isolate *Chlamydia trachomatis* even from people who are infected.
13. Discuss why chlamydia is more problematic and dangerous in females than males.
14. Babies born to females who acquire genital herpes late in a pregnancy are more at risk of contracting neonatal herpes than those whose mother had herpes prior to her pregnancy. Discuss why this occurs.
15. Discuss the relationship between HIV and AIDS.
16. Discuss why contact tracing is more important for STIs than some other infections.

9780170449090

CHAPTER 13 ACTIVITIES

ACTIVITY 13.1 Researching developments in contraception

The following is a list of research topics relating to the material in this chapter. In cooperation with your teacher, choose one of these topics to research and prepare a short talk to present to the rest of your class.

1. The search for a satisfactory oral contraceptive for males
2. The latest developments in the production of a contraceptive vaccine
3. Methods of birth control for less-developed countries
4. A brief history of birth control
5. The use of abortion as a means of birth control
6. The use of RU486 for medical abortion
7. The effectiveness of vasectomy reversal
8. Improvements in the detection of ovulation
9. The history of condom use and manufacture

ACTIVITY 13.2 Investigating the origin of HIV

Human immunodeficiency virus (HIV) was first identified as the cause of AIDS by French researchers in 1983, although it was not given its present name until 1986. Many hypotheses, some quite bizarre, were proposed to account for the origin of the virus. It is now generally accepted that HIV originated in monkeys or apes in sub-Saharan Africa and that sometime in the late 19th or early 20th century it was transferred to humans.

The identification of HIV as the cause of AIDS and the investigations about the origins of the virus are an interesting example of the way in which science works by the gradual accumulation of new knowledge as a result of painstaking research.

Using reliable sources of information, write a brief account of the history of the discovery of HIV and of its likely origins. A good place to start is the weblink 'The origin of HIV' on page 352.

ACTIVITY 13.3 Understanding the social consequences of vaccines for STIs

If effective vaccines for sexually transmitted infections (STIs) such as HIV, gonorrhoea, syphilis and chlamydia were widely available, who do you think should be vaccinated? What social and moral issues would be raised by the availability of such vaccines? Work in small groups, or organise a class debate, to argue the advantages and disadvantages of widespread vaccination against particular STIs. During the discussion, list all the advantages and disadvantages suggested.

9780170449090

CHAPTER 13 SUMMARY

- Contraception reduces the chance of pregnancy.
- Refraining from sexual intercourse, or abstinence, is the only contraceptive method that has no risk of pregnancy.
- Detecting ovulation via the rhythm method, the temperature method, the mucus method or a combination of these can allow couples to abstain from sexual intercourse before, during and after ovulation.
- During breastfeeding, ovulation is suppressed and so the chance of pregnancy is reduced. This is lactational amenorrhoea.
- Coitus interruptus is an unreliable contraceptive method that stops the semen entering the female by removing the penis prior to ejaculation.
- Mechanical barriers separate the sperm and the egg. These include:
  - condoms, which cover the penis
  - diaphragms, which cover the top of the vagina
  - cervical caps, which cover the entrance to the cervix
  - female condoms, which line the vagina.
- Spermicides may be used with most mechanical barriers to immobilise, and act as a barrier to, sperm.
- Hormonal contraceptive methods such as contraceptive pills, hormone implants and vaginal rings are effective methods that prevent ovulation by altering hormone levels.
- Intrauterine devices, or IUDs, are implanted in the uterus to release hormones or interfere with the movement of sperm.
- The morning-after pill uses progestogen to alter ovulation, stop the sperm reaching the egg and prevent implantation. This means that it is able to prevent pregnancy even after sexual intercourse as long as it is taken within five days.
- Sterilisation is a permanent method of contraception where the vas deferens or the uterine tubes are cut, a section is removed and the ends are tied. The procedure is called a vasectomy in males or tubal ligation in females.
- There are many reasons for choosing a particular method of contraception. This includes reliability, convenience, side effects, cost, permanence, religious beliefs and ethical concerns.
- Sexually transmitted infections are transmitted by close body contact, usually during sexual intercourse.
- Chlamydia is caused by the bacterium *Chlamydia trachomatis*. Those who show symptoms may develop infections of the urethra, epididymis or pelvic organs. In females, chlamydia may lead to infertility or an ectopic pregnancy. There is also the chance that it will be passed on to a foetus during birth. Chlamydia is diagnosed by a urine test or a swab of the affected areas. It can then be treated with antibiotics.
- Gonorrhoea is caused by the bacterium *Neisseria gonorrhoeae*. In males, it causes inflammation of the urethra, but this may spread to other organs if not treated. In females, it infects the urethra or cervix, and may spread to the uterine tubes and other organs. The inflammation may damage the oviduct to the point of infertility. Gonorrhoea can be treated with antibiotics.
- Syphilis is caused by the bacterium *Treponema pallidum*. The primary stage shows as chancres, usually on the sex organs. The secondary stage follows a few weeks to a year after the primary stage and shows as skin rashes, ulcerated mouth, fevers, and bone or eye disorders. The latent stage has no noticeable symptoms. The tertiary stage occurs between 5 years and 40 years after the initial infection. The symptoms during this stage are serious, such as heart disease.

9780170449090

- Genital herpes is caused by the herpes simplex virus, which produces blisters on the genital organs. Once a person is infected, the virus remains in the body for life. Therefore, the blisters can recur at any time.
- Genital warts are caused by the human papillomavirus (HPV). Some of the warts may be internal, and therefore it is possible for someone to pass the virus on unknowingly.
- Human immunodeficiency virus (HIV) is a retrovirus that infects T-lymphocytes. The infection goes through the stages of acute infection, chronic infection and then acquired immune deficiency syndrome (AIDS). The virus is transmitted through body fluids such as blood, seminal fluid and breast milk. HIV is diagnosed by a blood test to determine the antibody levels present or the viral load. While there is no cure for HIV, antiretroviral drugs are able to inhibit the reproductive cycle of the virus and delay its progression.
- STIs can be prevented by practising safe sex.

9780170449090

CHAPTER 13 GLOSSARY

Abstinence Refraining from sexual intercourse

Acquired immune deficiency syndrome (AIDS) An extremely serious and often fatal disease caused by infection with human immunodeficiency virus (HIV) that damages the immune system so that the individual becomes susceptible to infection and to some forms of cancer

Castration The removal of the testes

Cervical cap A thin rubber cap that is fitted across the cervix before sexual intercourse; it prevents sperm entering the uterus

Chancres The initial stage of a syphilis infection; small sores that appear on the penis, labia of the vagina, and sometimes other parts of the body

Chlamydia One of the most common sexually transmitted infections; transmitted by the *Chlamydia trachomatis* bacterium; results in inflammation of the urethra in males and of the uterus and uterine tubes in females

Coitus interruptus A method of birth control that depends on the withdrawal of the penis from the vagina just before ejaculation

Combined pill A contraceptive pill that contains oestrogen and progesterone

Condom A thin sheath of latex that is rolled on to the erect penis before sexual intercourse to prevent sperm from entering the vagina; a means of contraception

Contact tracing The process of identifying the relevant contacts of a person with an infectious disease to ensure that they are aware of their exposure to the infection; for sexually transmitted infections (STIs), relevant contacts include those with whom the infected person has had sex during the infectious period

Contraception The prevention of conception; birth control

Diaphragm A thin rubber cap that is fitted across the vagina before sexual intercourse to prevent sperm from entering the uterus

Ectopic pregnancy A pregnancy in which the embryo implants at a site other than in the lining of the uterus, most frequently in one of the uterine tubes

Epididymitis Inflammation or infection of the epididymis; results in pain and swelling of the epididymis

Femidom A female condom; a lubricated polyurethane sheath that lines the vagina

Genital herpes A viral infection of the genital region caused by the herpes simplex type 2 virus; results in persistent and painful blisters

Genital warts Warts in the genital region transmitted during sexual intercourse and caused by the human papillomavirus

Gonorrhoea One of the most common sexually transmitted infections; transmitted by the bacterium *Neisseria gonorrhoeae* during sexual intercourse; infects the mucous membranes of the reproductive organs; commonly referred to as 'the clap'

Herpes simplex virus Causes genital herpes, a viral infection of the genital region

Human immunodeficiency virus (HIV) A virus that is transmitted by infected body fluids such as blood and semen, and causes progressive damage to the body's immune system; frequently transmitted during unprotected sexual intercourse

Human papillomavirus (HPV) A highly contagious virus spread through close contact. Some types of HPV cause genital warts; nearly a third of all the known types of HPV can be transmitted through sexual contact

Hysterectomy The complete or partial removal of the uterus

Implanon NXT A plastic rod that is implanted under the skin to slowly release progestogen to prevent pregnancy

Incubation period The time between infection and the appearance of the symptoms of a disease

Intrauterine device (IUD) A plastic or metallic device inserted into the uterus to prevent conception

9780170449090

Lactational amenorrhoea Temporary infertility that follows the birth of a child

Latent stage The dormant or hidden stage of the course of a disease or infection; one of the stages of a syphilis infection

Mini pill A contraceptive pill that contains only progestogen

Morning-after pill Emergency contraceptive pill, taken after sexual intercourse

Mucus A thick fluid secreted by mucous glands and mucous membranes

Mucus method A method of contraception whereby the consistency of the cervical mucus is used to estimate when ovulation is occurring

Non-specific urethritis (NSU) A sexually transmitted infection usually caused by the *Chlamydia trachomatis* bacterium

NuvaRing A soft plastic vaginal ring that contains oestrogen and progestogen

Oophorectomy The surgical removal of the ovaries

Pandemic An infection that has spread to all parts of the world.

Pelvic inflammatory disease (PID) A bacterial infection of the pelvic organs of a female, especially of the uterus and uterine tubes

Periodic abstinence A method of contraception based on abstaining from sexual intercourse when fertilisation is most likely

Pubic lice Insects that infest the genital area causing intense itching; usually transmitted by sexual intercourse

Retrovirus A virus that contains RNA instead of the more usual DNA

Rhythm method A method of contraception in which sexual intercourse is avoided around the time that ovulation is likely to occur

Safe sex Sexual activity in which precautions are taken that reduce the risk of getting a sexually transmitted infection (STI) or an unplanned pregnancy

Scabies A mite that burrows into the skin causing intense itching, frequently in the genital area; often transmitted by sexual intercourse

Sexually transmitted disease (STD) *see* sexually transmitted infection

Sexually transmitted infection (STI) An infection transmitted by direct sexual contact, usually through sexual intercourse; formerly referred to as sexually transmitted disease (STD)

Spermicide Chemical that immobilises or kills sperm

Symptothermal method A method of contraception that combines the rhythm, temperature and mucus methods

Syphilis A sexually transmitted infection caused by the bacterium *Treponema pallidum*; often referred to as 'the pox'

Temperature method A variation of the rhythm method of contraception, where the woman's body temperature is used to estimate when ovulation is occurring

Trichomoniasis An infection caused by the protozoan *Trichomonas vaginalis* that results in inflammation of the mucous membranes of the vagina in women and the urethra in men

Tubal ligation Female sterilisation; the removal of a small piece, or the clamping off, of each uterine tube

Vasectomy Male sterilisation; the removal of a small piece of each vas deferens

9780170449090

CHAPTER 13 REVIEW QUESTIONS

Recall

1 a Define 'contraception'.
  b List the methods of contraception available to both a man and a woman.
  c Draw up a table comparing each of the methods of contraception discussed in this chapter. In your table, include columns for reliability, advantages and disadvantages.

2 a Define 'sexually transmitted infection'.
  b What types of organisms can cause STIs?

3 a Outline the principle behind the rhythm method of birth control.
  b Describe two ways in which the time of ovulation can be detected.
  c List advantages and disadvantages of the various 'safe period' methods as a means of birth control.

4 Describe the advantages of diaphragms.

5 a Briefly outline the way in which hormonal methods of contraception work in females.
  b List the various ways in which these hormonal methods can be administered.
  c List the disadvantages of hormonal contraceptives.

6 Which parts of the body are affected by infection with chlamydia in:
  a males?
  b females?

7 Use a table to compare the causes, symptoms and treatment for gonorrhoea, syphilis, HIV, genital herpes and chlamydia.

8 HIV is a serious infection with significant ramifications for individuals and communities.
  a Explain what is meant by 'viral load', and describe how viral load can be used as a test for HIV infection.
  b Outline the stages of an infection with human immunodeficiency virus (HIV) that eventually develops into AIDS.
  c What complications are associated with AIDS?
  d Briefly outline the way in which HIV may be spread from person to person.
  e What preventive measures are available to reduce the risk of infection by HIV?

9 Explain how an infant could be infected with:
  a syphilis
  b gonorrhoea.

Explain

10 Explain the disadvantages of coitus interruptus as a method of birth control.

11 Explain why some couples prefer not to use diaphragms or cervical caps as their method of birth control.

12 Explain how breastfeeding can prevent pregnancy.

13 Explain how IUDs prevent pregnancy.

14 Explain how chlamydial infection can lead to infertility in women.

Apply

15 Compare and contrast vasectomy and tubal ligation.

16 Discuss the factors that a person should consider before having a vasectomy or a tubal ligation.

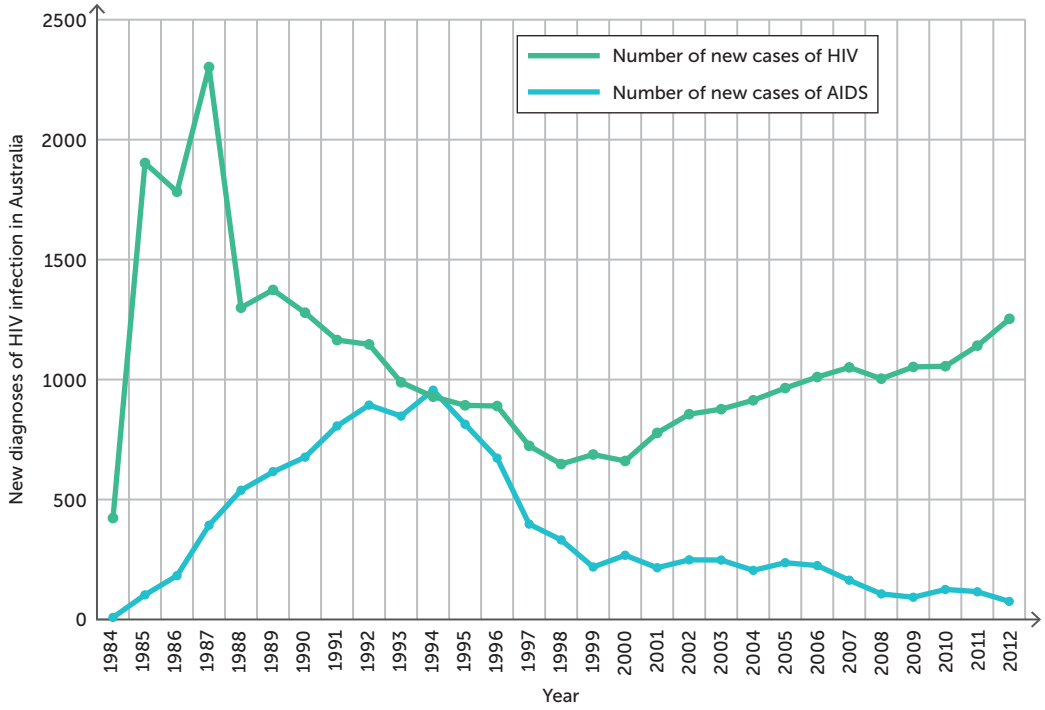
17 'People don't die from HIV.' Discuss this statement.

18 How do sexually transmitted infections differ from other communicable diseases? List as many differences as possible.

9780170449090

19 The graph below shows the number of new HIV and AIDS cases in Australia from 1984 to 2012. Suggest why the number of cases of AIDS has declined but there has been a less significant decline in new cases of HIV infection. (In 2019, the number of new diagnoses of HIV was 937.) Would you expect this same trend worldwide? Explain your answer.

20 Some people refer to STIs as 'social' diseases. Explain how this term may have arisen.



Extend

21 Explain why many millions of dollars are spent annually on research into birth-control techniques. Give as many reasons as you can for the commercial and social importance of birth control.

22 Rising population is a major problem in many countries. High birth rates occur in less-developed countries, rather than in developed countries such as Australia. Of the contraceptive measures described in this chapter, which do you think would be most suitable for use in a less-developed country? Which would be least suitable? Give reasons for your answers.

23 Which partner in a sexual relationship should have the responsibility for contraception? How should this decision be arrived at by the couple? Write a short essay to argue your case.

24 In Australia during 1988, nine people died from syphilis. Eight of these were people over the age of 65 and one was a child in her first year of life. From your knowledge of the progress of this disease, account for the marked age variation in these statistics.

25 The incidence of syphilis in Australia remained relatively constant from 2004 to 2017 (see Figure 13.17), whereas

9780170449090

the incidence of gonorrhoea increased greatly over the same period. Suggest as many reasons as you can to account for this difference in the incidence of the two diseases.

26 The infections discussed in this chapter are those that are commonly considered to be STIs. There are a number of other diseases that can be transmitted sexually, but that is not their only mode of transmission. Two such conditions are hepatitis B and molluscum contagiosum. For each of these infections, find out:

- **a** the infective agent that causes the disease
- **b** how the disease is transmitted – sexually and by other means
- **c** the symptoms of the disease
- **d** the treatment for the disease.

9780170449090

14 TECHNOLOGIES ARE AVAILABLE TO ASSIST IN REPRODUCTION

UNIT 2 CONTENT

SCIENCE INQUIRY SKILLS

» interpret a range of scientific and media texts, and evaluate processes, claims and conclusions by considering the quality of available evidence; and use reasoning to construct scientific arguments

SCIENCE AS A HUMAN ENDEAVOUR

» the use of genetic profiling and genetic screening of adults and embryos have implicit ethical considerations

» greater understanding of the menstrual cycle, conception and implantation has produced improved methods of the establishment of a pregnancy, along with advancements in contraceptive methods; both have ethical considerations

SCIENCE UNDERSTANDING

Human reproduction

» there are a variety of assisted reproductive technologies to help overcome infertility problems, but each has its limitations, risks and benefits

» there are a range of techniques available to genetically screen embryos before implantation or during early development, including blood tests, amniocentesis and chorionic villi sampling

Source: School Curriculum and Standards Authority, Government of Western Australia

9780170449090

Unfortunately, sometimes there are difficulties in establishing pregnancy. Over time, our understanding of the process has increased immensely. This has allowed the development of more effective methods of testing, establishing and sustaining pregnancy. However, as with any advancement, there are various factors and regulations that must be considered in regard to these technologies.

14.1 TREATMENT OF INFERTILITY

Infertility is defined as being unable to achieve pregnancy despite frequent unprotected sex over the period of at least a year. In Australia, infertility affects approximately one in six couples.

Causes of infertility

About 40% of infertility is due to problems with the sperm. Another 40% is due to problems in the female reproductive system, while the remaining 20% is due to a combination of male and female factors.

Sperm production

For sperm to be able to fertilise an egg, it must be:

- produced in sufficient quantities
- able to move in a forward direction
- able to penetrate the corona radiata and zona pellucida.

This means that there needs to be a high number of sperm produced with a correct structure. Problems with any of these qualities will greatly reduce the chance of a pregnancy.

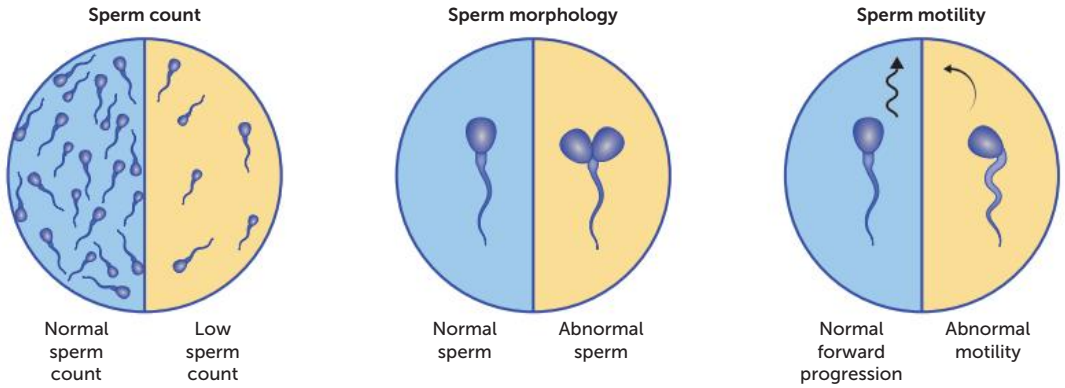


FIGURE 14.1 Sperm factors that affect fertility

Other factors affecting male fertility

There are several other factors that may result in sperm being unable to fertilise an egg.

- Semen, and the sperm it contains, may flow into the bladder, rather than out the urethra.
- The male's immune system may develop antibodies for their own sperm, altering them and reducing their effectiveness.
- Blockages may occur in the male's reproductive tract, especially in the vas deferens, which may stop the sperm leaving the testes.
- Hormonal imbalances can affect sex drive and the production of sperm.

Sperm quality
This website offers a detailed explanation of sperm and their impact on fertility.

Alamy Stock Photo/Cultura Creative (RF)

FIGURE 14.2 Sperm morphology, or structure, is a factor relating to fertility. Sperm with bent tails are unable to move, making it more difficult for them to reach the egg

Ovulation

As females get older, the number of healthy eggs remaining decreases. This greatly reduces the chance of pregnancy, especially after the age of 36.

Polycystic ovarian syndrome (PCOS) is another factor affecting the fertility of females. PCOS is a hormonal condition where the ovaries contain many partially formed follicles that fail to mature. This means that the eggs are not released, and therefore cannot be fertilised.

Hyperprolactinemia, which means high levels of the hormone prolactin, may occur in people with pituitary tumours, hypothyroidism and PCOS. The high level of prolactin results in intermittent or a lack of ovulation.

Once a female has gone through menopause, she will no longer be ovulating. Some females experience menopause before the age of 40 and therefore will be unable to get pregnant from this time. This is called early menopause, or primary ovarian insufficiency.

Cancer treatments may also cause primary ovarian insufficiency, and hence infertility. In women younger than 30, this may be temporary, and they may become fertile again after treatment. The older a woman is, the less likely it is that her fertility will return.

Fertility and a woman's age
This website contains a video about the effect of age on a woman's eggs.

Problems with ovulation
This Australian website has a video discussing problems with ovulation.

Other factors affecting female fertility

Endometriosis affects approximately 10% of women. It is a painful condition where the cells of the endometrium grow outside the uterus. The resulting scar tissue or distortion of the uterine tubes can affect fertility by blocking the egg's release or pathway through to the uterus.

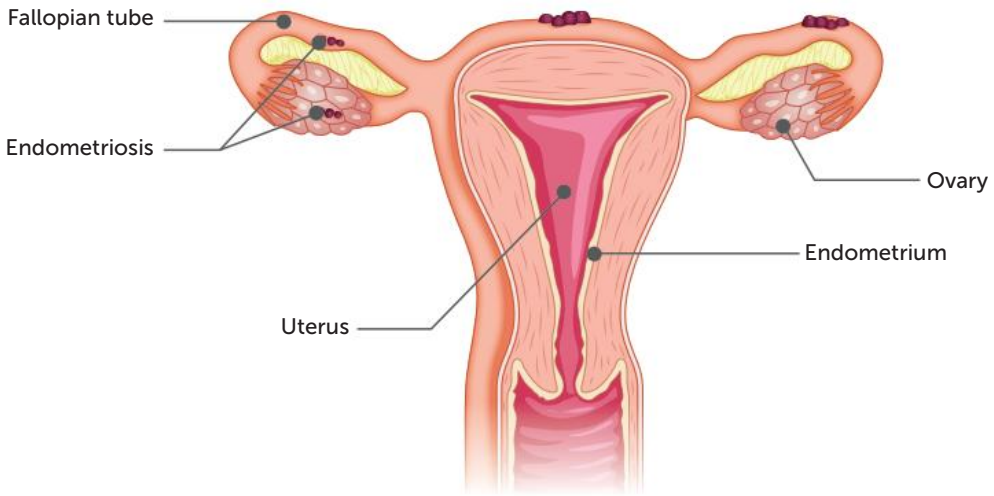


FIGURE 14.3 Endometriosis occurs when cells of the endometrium grow outside the uterus

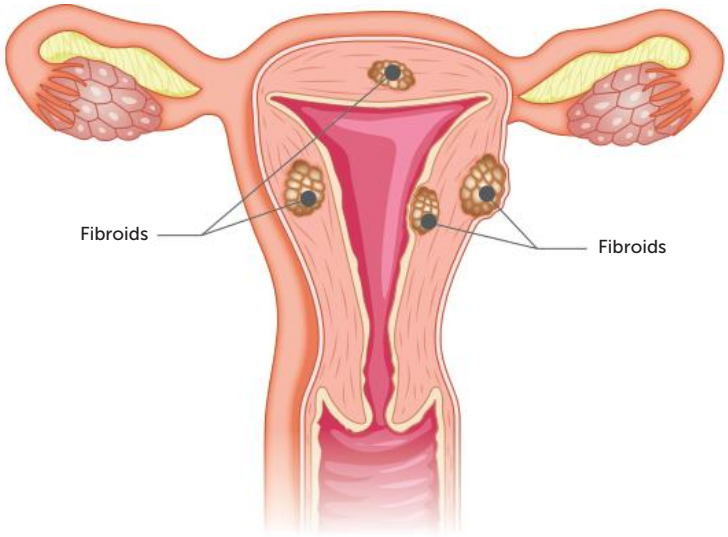


FIGURE 14.4 Fibroids are benign growths in the uterus

The presence of fibroids may also affect fertility. **Fibroids** are benign growths in the muscular part of the uterus. While they are very common, they only affect fertility if their location distorts the uterine cavity or blocks the uterine tubes.

Blockages of the uterine tubes will stop the egg passing through to the uterus. This can occur following infections, such as gonorrhoea or chlamydia, or damage – for example, due to ectopic pregnancies.

Problems with the menstrual cycle can also affect fertility as it influences implantation of the blastocyst. Hormonal imbalances can reduce the development of the endometrium and its maintenance.

Key concept

Any factor that limits the possibility of a sperm fertilising an egg, and the resulting embryo implanting, will reduce the chances of pregnancy.

Infertility treatments that allow unassisted fertilisation

The method of treating infertility depends on the reason for the issue. In some instances, it is possible to correct the problem so that the couple can conceive naturally. In other situations this is not possible, and so techniques are used to assist fertilisation and the maintenance of pregnancy.

Surgery

Microsurgery can be used to solve some problems of infertility. Blocked uterine tubes and sperm ducts can be opened, and fibroids or endometriosis can be removed.

Ovulation tracking

It is possible to identify the time when a female is most fertile through a series of blood tests. There is a surge in luteinising hormone prior to ovulation. The level of the hormone can be monitored through blood tests so that a couple knows when they are most likely to conceive. Sperm remain viable for two to three days in the uterine tubes, while the egg only survives for 24 hours. Therefore, the highest chance of conception occurs by insemination prior to ovulation. If a couple tracks the ovulation of the female over a number of cycles, it is possible to identify the most effective time to have sexual intercourse.

9780170449090

Ovulation induction

In some cases, infertility is due to problems with ovulation resulting from a low level of hormones. These problems may be solved by medications used to correct the problem. Two types of drugs are used, both of which use follicle-stimulating hormone (FSH) to induce the development of follicles.

- Clomiphene stimulates the body to make more FSH.
- Hormone injections of FSH increase the blood levels of the hormone.

Another hormone, human chorionic gonadotrophin (hCG), may also be used to trigger ovulation once the follicle has matured.

Females affected by hyperprolactinemia will need to lower their prolactin levels so that ovulation can occur. Correction of the underlying cause is an important consideration. Oral medication is also available to restore normal levels of prolactin and, hence, ovulation.

Artificial insemination

Artificial insemination, or **intrauterine insemination (IUI)**, is a process where sperm is released into the uterus by a catheter being inserted through the cervix. The sperm sample is then injected through the catheter. From the uterus, the sperm are able to move naturally through the uterine tubes where they may fertilise an egg. IUI increases the chance of pregnancy for affected couples by increasing the number of sperm that reach the uterine tubes.

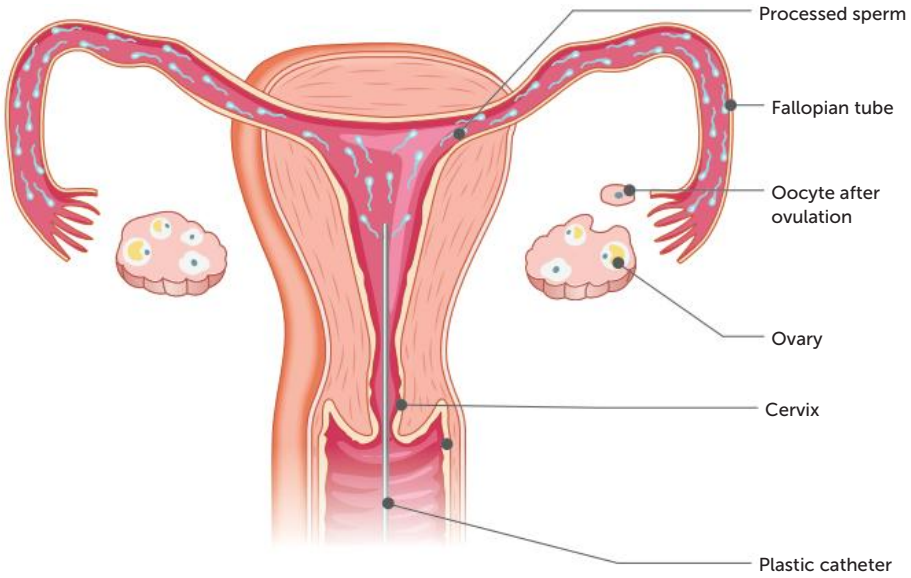


FIGURE 14.5 Intrauterine insemination

Artificial insemination has the advantage of allowing control over the sperm being inseminated. The sperm used may be the male partner's or from a donor male. In both instances, the sperm is collected, analysed, processed and concentrated. This ensures that only high-quality sperm are used. It is also possible for the sperm to be collected at an earlier time and frozen for storage. The sperm is then thawed under controlled conditions prior to insemination.

There are a range of circumstances that would benefit from IUI. These include:

- males with a low sperm count or decreased motility
- ejaculation dysfunctions
- sperm stored and frozen due to a male's absence or needing to undergo treatment such as chemotherapy or radiotherapy
- females with cervical scarring
- hostile cervical mucus
- same sex couples
- single females.

9780170449090

The success rate for suitable females achieving a pregnancy using IUI is approximately 10–20% per cycle. Over three to six cycles, there is on average an 80% chance of getting pregnant.

The success rate for couples with no fertility issues ranges from 96%, on average, if the woman is under 25 years of age to 78% if the woman is 37 years old. Therefore, the success rate using IUI is relatively high.

Key concept

Some treatments, such as surgery, ovulation tracking, ovulation induction and artificial insemination, may overcome a fertility issue and allow the sperm to fertilise an egg naturally in the uterine tube.

Assisted fertilisation

It is not always possible for natural fertilisation to occur. In these cases, there are various **assisted reproductive technologies** that can be considered.

Gamete intrafallopian transfer

Gamete intrafallopian transfer (GIFT) is a procedure that can be performed when there are normal uterine (or fallopian) tubes and adequate sperm. It involves the following steps:

1. Hormonal treatment is used to stimulate the female to produce more than one egg.
2. The sperm and egg are collected and analysed.
3. The sperm and egg are mixed together in the laboratory.
4. The sperm and egg mixture is injected into the woman's uterine tubes during laparoscopic surgery.

It is hoped that the sperm will then fertilise the egg naturally and move down the uterine tube before implanting in the uterus.

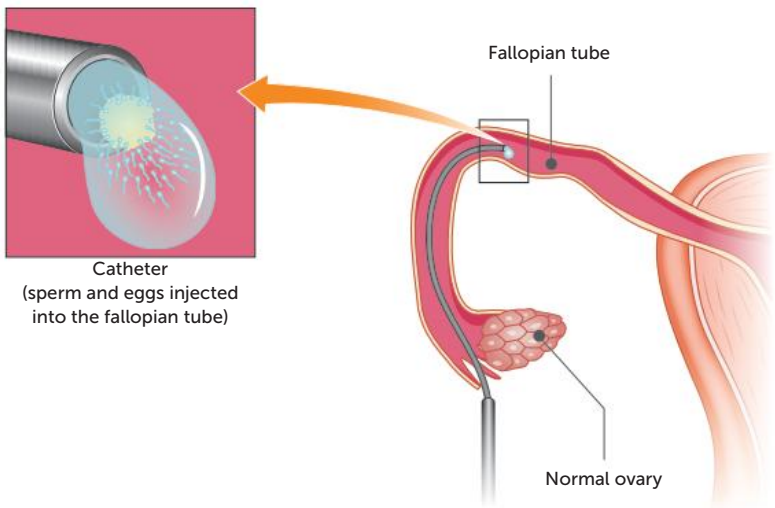


FIGURE 14.6 Gamete intrafallopian transfer (GIFT) involves sperm and eggs being injected into the uterine (or fallopian) tubes

As advancements have been made in other areas of assisted reproductive technologies, the frequency of GIFT procedures has decreased. GIFT has the disadvantages of having a lower pregnancy rate and requiring surgery. However, as fertilisation occurs naturally, this method is preferred by some couples who are unable to use other techniques due to religious or ethical reasons.

In vitro fertilisation

In vitro fertilisation (IVF) has been used since the first 'test tube baby' was born on 25 July 1978. It is used to overcome a range of fertility issues, such as blocked uterine tubes, ovulation disorders, endometriosis, fibroids, low sperm quality or production, and unexplained infertility.

During IVF, hormonal treatment is used to:

- stimulate the ovaries so that multiple follicles develop
- control ovulation
- prepare the uterine lining.

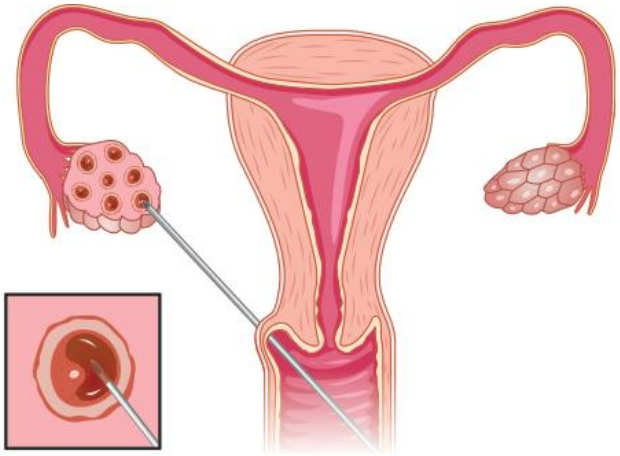


FIGURE 14.7 Egg retrieval for IVF

TED-Ed: How in vitro fertilization works

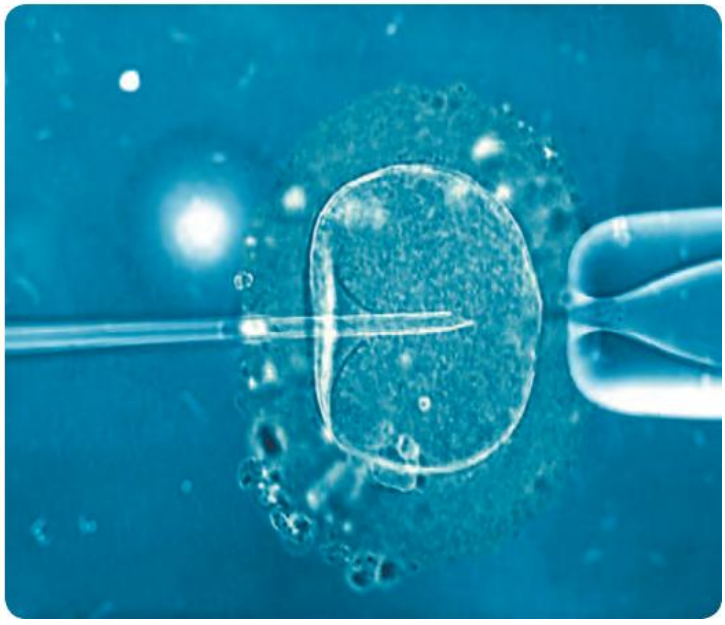
A series of blood tests and ultrasounds are used to monitor the development of the follicles. When the eggs are mature, they are collected by a needle passing through the vagina to the ovaries.

Following their retrieval, the eggs are mixed with sperm in a suitable environment at 37°C to maximise their chances of fertilisation and development. Approximately two to six days after collection, an embryo is inserted into the uterus via a catheter passed through the cervix.

In Australia, there are regulations regarding the number of embryos transferred per cycle to limit the risk of multiple pregnancies. This means that there may be unused embryos. These are able to be frozen for use in future cycles.

Intracytoplasmic sperm injection

If there is a problem with the sperm, it is likely that it will not be able to fertilise the egg unassisted. This means that IVF has a low chance of success. **Intracytoplasmic sperm injection (ICSI)** is very similar to IVF. Once the eggs have been collected, they are examined to ensure that they are suitable. A sperm is then injected into the egg, achieving fertilisation. The resulting cells are then monitored, and any that are developing normally are able to be inseminated.



Science Photo Library/Zephyr

FIGURE 14.8 Intracytoplasmic sperm injection. The photograph shows a sperm being injected into an egg

One concern regarding the use of ICSI is that the reason for the male infertility may be a genetic disorder. Using ICSI to overcome the abnormality means that the genetic defect may be passed on to the offspring. For this reason, it is recommended that children born from ICSI are examined by a paediatrician. It is also possible for genetic testing to be done prior to undertaking ICSI, and if the male is found to have the genetic defect, other options may be considered.

Surgical sperm retrieval

Some men are unable to ejaculate, or very low numbers of sperm are released. In these instances, sperm may be collected during surgery to be used in IVF and ICSI. The surgery may be performed under local or general anaesthetic. In both cases, a needle is used to collect sperm from the epididymis or testis.

Key concept

Methods such as gamete intrafallopian transfer, in vitro fertilisation, intracytoplasmic sperm injection and surgical sperm retrieval increase the chances of conception with some fertility issues.

Other options for pregnancy

Unfortunately, it is not always possible for a couple to use their own egg and sperm, or for the female to carry the child.

Donor gametes or embryos

It is possible to use eggs, sperm or embryos from donors to achieve a pregnancy. The donor may be known to the couple, or anonymous. In Australia, there are strict regulations regarding donors, including counselling for both donors and recipients, cooling-off periods and health screening.

Surrogacy

If a female is unable to conceive or carry a baby, **surrogacy** is an option that they may consider. With this arrangement, another woman carries the child for the duration of the pregnancy and then gives the child to the couple to raise as their own.

There are very strict laws regarding surrogacy in Australia. While these vary between states, it is not legal for the surrogate, or birth, mother to be paid beyond expenses arising from the pregnancy. Additionally, the agreement between the birth mother and the parents is not legally binding. This means that the birth mother has the right to change her mind and keep the child.

Traditionally, a surrogate pregnancy was achieved by artificial insemination of the father's sperm. This means that the surrogate mother is genetically related to the child. An alternate method is gestational surrogacy, where IVF is used to produce an embryo that is then inserted into the surrogate. This is more common in Australia.

Key concept

Donor sperm, egg or embryo are options for people whose own sperm and eggs are unable to be used to conceive, while surrogacy may be an option for people who are unable to conceive or carry a child.

14.1 Reproductive technologies

Other considerations regarding fertility treatments

As with many advancements, there are factors that must be considered and regulated with fertility treatments.

Frozen embryos

Embryos that are produced but not used during an IVF cycle are typically frozen. This process is now highly developed, with the embryos, often in the blastocyst stage, being cooled to –196°C and stored in liquid nitrogen tanks. The embryos stop developing, and remain in the same stage until they are thawed.

Alamy Stock Photo/JOMWASCHARA KOMVORN

FIGURE 14.9 Frozen embryos are stored in liquid nitrogen tanks

In 2019, it was estimated that there were more than 100 000 frozen embryos stored in Australia. While the embryos can be kept indefinitely, there are regulations guiding this. They vary among the states; however, five years is the typical limit. But this can often be extended through consultation with specialists.

When the embryos are no longer needed by a couple, a decision must be made regarding what to do with them. The options are to:

- dispose of the embryos
- donate them to other people/couples
- donate them for research.

The outcome of embryos is a serious ethical and moral consideration. For this reason, there are many legalities and guidelines relating to this area of assisted reproductive technology. There are also various choices available for those involved, to accommodate different religious and personal beliefs and values.

Religious beliefs

Religions differ in their acceptance of assisted reproduction. Some concerns are: the involvement of a third party in the process of fertilisation; the fate of excess embryos; and the separation of procreation and sexual function. The decision regarding whether or not to use assisted reproductive technologies, and which methods to use, is an individual one, with religious beliefs being one guiding factor.

Cost

Assisted reproductive technologies are very expensive. Some costs involved are:

- specialist doctor consultations
- initial tests and screenings
- medications
- fertility treatments such as IVF and ICSI
- surgery and/or hospital costs
- access to the donor program
- frozen sperm, egg or embryo storage.

While Medicare rebates are available for many treatments, the out-of-pocket expenses per cycle are still very high. Most couples require multiple cycles to fall pregnant; therefore, assisted reproduction becomes a very expensive option. For example, one clinic in Australia states that the cost of IVF is $9828 per cycle, with an out-of-pocket expense of $4991 (IVF Australia, 2020).

Activity 14.1
Should we use assisted reproductive technologies?

Questions 14.1

RECALL KNOWLEDGE

1 Define 'infertility'.
2 List the reasons that a male may be infertile.
3 Describe polycystic ovarian syndrome.
4 Define 'endometriosis' and describe why it affects fertility.
5 Gonorrhoea and chlamydia may both result in scarring of the uterine tubes. How does this affect fertility?
6 Explain how ovulation tracking can increase the chances of conception.
7 What hormone is used to stimulate the development of follicles in the ovary? Explain how this increases the chances of pregnancy.
8 Describe a situation where it is unlikely that artificial insemination would be successful.
9 What does 'GIFT' stand for?
10 Describe the steps involved in IVF.
11 List five factors that a person or couple would need to think about when considering using assisted reproductive technologies.
12 List four possible outcomes for frozen embryos.

APPLY KNOWLEDGE

13 Explain why a high percentage of sperm with a bent tail reduces the chances of conception.
14 Explain why a female is less likely to conceive when she is 40 years old than when she was 25 years old.
15 List two similarities and two differences between endometriosis and fibroids.
16 The highest chance of conception occurs when insemination is two to three days prior to ovulation. Use your understanding of human reproduction to explain this observation.
17 Intrauterine insemination can have a higher success rate than natural insemination for some couples. Explain why this happens.
18 To get pregnant, some couples may consider IVF, but not ICSI. Suggest why they would consider IVF acceptable, but not ICSI.
19 Gestational surrogacy is more common than traditional surrogacy. Suggest a reason why a:
   a heterosexual couple might use traditional surrogacy
   b heterosexual couple might use gestational surrogacy
   c homosexual couple might use traditional surrogacy.

14.2 DIAGNOSIS OF FOETAL HEALTH

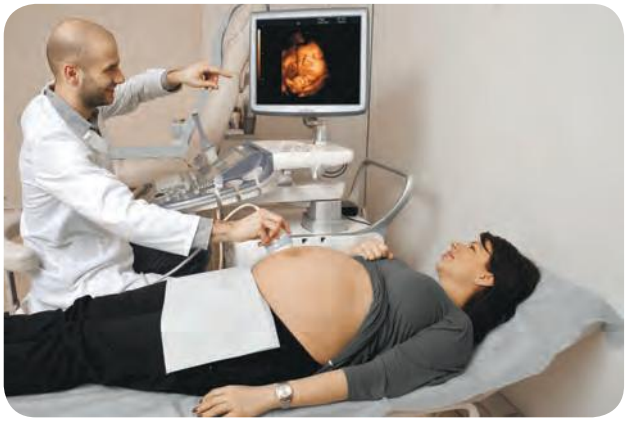
To maintain a healthy pregnancy, a woman needs to ensure that she has regular medical checks with her doctor or other heath-care professional. In this way, the health of the foetus can be monitored. If there is concern at any stage, a range of technologies is available for testing the foetus. Two of these techniques allow an image of the foetus to be seen, whereas others provide an analysis of the chromosomes or of some of the chemicals from the foetus. Table 14.1 lists the techniques available and the year in which general use of the test began. In addition, counselling is available for couples concerned about the risks of producing a baby with a birth defect.

TABLE 14.1 History of foetal diagnosis

TECHNIQUE	YEAR WHEN GENERAL USE BEGAN
Ultrasound	1957
Amniocentesis	1970
Amniocentesis with ultrasound guidance	1972
Fetoscopy	1974
Chorionic villus sampling	1983
Foetal blood sampling	1983

Ultrasound

Ultrasound uses inaudible, high-frequency sound waves to produce an image of the foetus. A probe is placed on the abdomen of the pregnant woman, and the sound waves are reflected by the foetal tissues to obtain a visual 'echo' of what is inside the uterus. The doctor feeds these reflected sounds, or echoes, into a computer to produce a screen image of the foetus for study.



Shutterstock.com/bart78

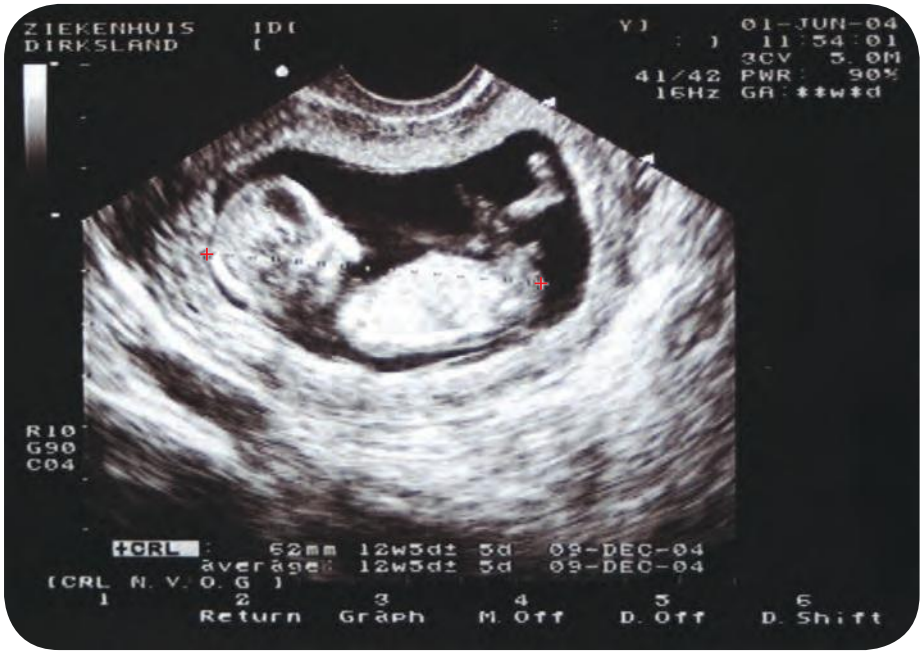
FIGURE 14.10 A pregnant woman being examined by ultrasound

The images produced by ultrasounds can be used to monitor the growth and development of the foetus. The information that can be gained from ultrasounds includes:

- confirming pregnancy
- estimating the stage of pregnancy
- determining the number of foetuses
- identifying abnormalities of the cervix or uterus
- monitoring the growth of the foetus
- determining the gender of the foetus
- evaluating the anatomy of the foetus
- genetic screening
- studying the placenta and amniotic fluid
- identifying birth defects
- determining the position of the foetus.

9780170449090

While foetal ultrasound is a safe technique for gaining valuable information, it has its limitations. It cannot diagnose all abnormalities and, if an abnormality is detected, further tests may be required for more specific information.



Dreamstime.com/Pilene

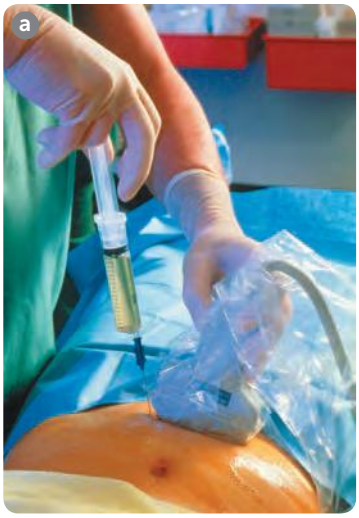
FIGURE 14.11
Ultrasound image of a 12-week-old foetus. The distance between the two red crosses on this image is known as the crown–rump length

Genetic analysis

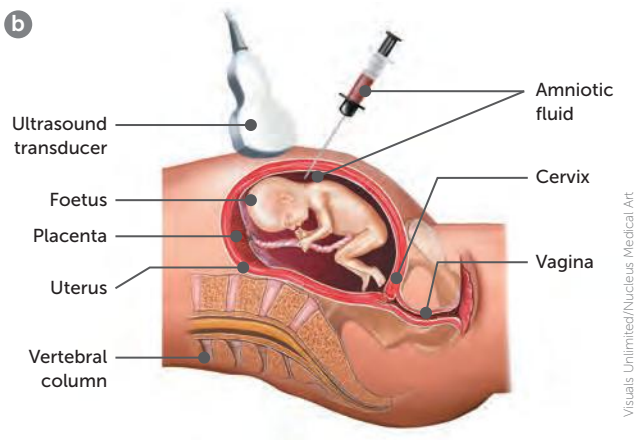
Foetal cells can be obtained for analysis using either amniocentesis or chorionic villus sampling. These tests can be used to examine the foetus's chromosomes to detect defective, missing or additional chromosomes.

Amniocentesis

Amniocentesis is carried out between the 16th and 20th weeks of pregnancy, by which time the foetus is floating in about 130 mL of amniotic fluid. It involves using an ultrasound to guide a needle through the abdominal wall into the amniotic cavity. Approximately 10–20 mL of the fluid is removed. Living cells from the foetus are floating in the fluid. These cells can be examined for biochemical defects and for abnormalities in the number of chromosomes or in the chromosome structure.



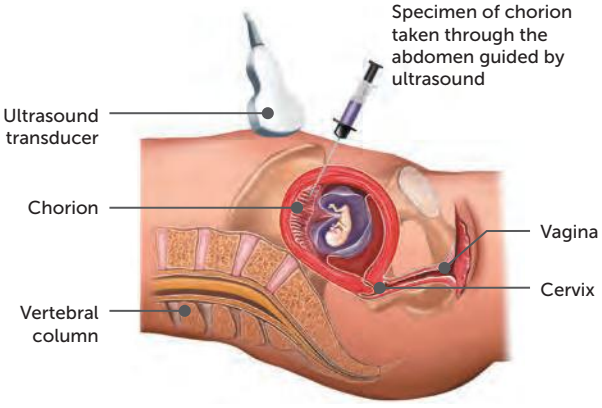
Science Photo Library/SPL/BSIP Laurent



Visuals Unlimited/Nucleus Medical Art

FIGURE 14.12
a A doctor performs amniocentesis on a pregnant woman; **b** Amniocentesis involves taking a sample of amniotic fluid (which carries foetal cells) with a needle

Amniocentesis involves a small risk of infection, miscarriage or damage to the baby. It is therefore only performed on women who are thought to be at higher risk of delivering a child with a birth defect. Some of the disorders that can be detected by this procedure are Down syndrome, cystic fibrosis, neural tube defects such as spina bifida, and a wide range of genetic disorders, including phenylketonuria, Tay-Sachs disease, Duchenne muscular dystrophy and sickle cell disease.



Visuals Unlimited/Nucleus Medical Art

FIGURE 14.13 In chorionic villus sampling, a specimen of foetal cells is taken from the chorion

Chorionic villus sampling

Chorionic villus sampling (CVS) obtains a specimen of foetal cells from the chorion, one of the foetal membranes. The cells are then examined in a similar way to those gained by amniocentesis. CVS has an advantage over amniocentesis, however, in that testing can take place at 9–19 weeks of pregnancy. In addition, the foetal tissue gained through CVS can be tested more quickly than the specimen of amniotic fluid, thus reducing the time between the testing procedure and examination of the results. This is especially important if a birth defect is involved that may require termination of the pregnancy. A disadvantage of CVS is that the risk of miscarriage following the procedure is 2%. CVS can be used to detect genetic disorders and biochemical abnormalities, but it cannot diagnose spina bifida.

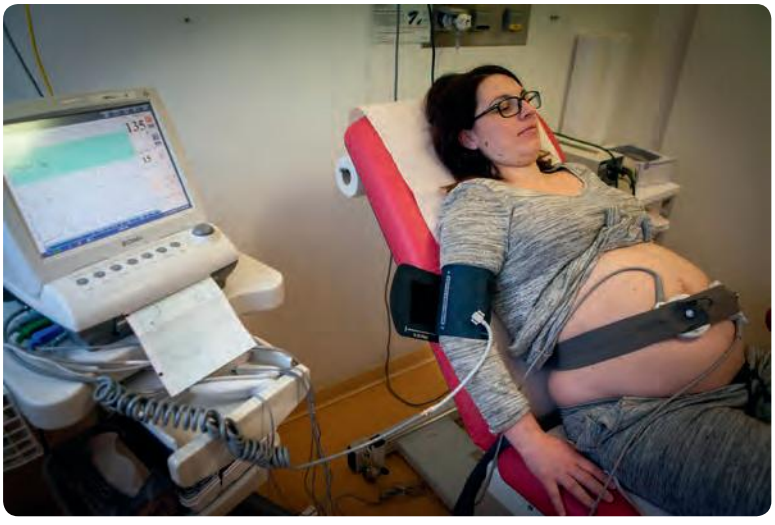
Blood tests

A blood test of the mother's blood is a non-invasive prenatal test that is possible due to some of the baby's DNA passing into the mother's blood. This is a screening test, not a diagnostic one. This means that it is able to identify if there is an increased chance of the baby having a certain disorder; it does not confirm whether the baby definitely has the disorder.

The blood tests are able to screen for:

- Down syndrome (also called trisomy 21)
- Edwards syndrome (trisomy 18)
- Patau syndrome (trisomy 13)
- Turner syndrome.

The blood tests are available from approximately 10 weeks of gestation. If they show an increased chance of an abnormality, an amniocentesis or chorionic villus sampling would be recommended to confirm the diagnosis.



Alamy Stock Photo/BSIP SA

FIGURE 14.14 Foetal monitoring allows continuous evaluation of a foetus. It can give early warning of foetal distress and allows precise management of labour

Foetal monitoring

Foetal monitoring is the regular recording of a baby's heart rate in order to detect indicators of stress. This monitoring usually takes place during labour and birth using ultrasound and electrocardiography. **Electrocardiography** is a procedure for recording electrical changes in the heart. The record, which is called an **electrocardiogram (ECG)**, shows the series of waves that relate to the electrical impulses that occur during each beat of the heart. The results are printed on paper or displayed on a monitor.

The aim of foetal monitoring is to identify any risk of injury to the foetus so that appropriate action can be taken. A foetal monitor may be used during labour and birth to record the baby's heart rate, and sometimes the mother's contractions. A detailed foetal heart rate analysis enables medical staff to check whether there is any risk of oxygen deficiency occurring. Oxygen deficiency during birth may result in brain damage, or even a stillbirth.

Fetoscopy

A **fetoscope** is an instrument used to gain information about a foetus in the uterus. There are two types of fetoscopes: a stethoscope that listens to the foetu's heartbeat; and a fibre-optic scope that looks directly at the foetus through a small, telescope-like instrument with a diameter about the size of a large hypodermic needle. The fibre-optic fetoscope is introduced into the uterus through the abdominal wall. Examination of the outward appearance of the foetus may enable the detection of such conditions as cleft lip and palate, missing or abnormal ears, deformed or absent limbs, and spinal abnormalities, including spina bifida. If detection takes place early in the pregnancy, a decision about termination can then be made. Fetoscopy is also used during foetal surgery.

Fetoscopy is a risky, difficult procedure that is only performed by a specialist doctor, usually after an ultrasound has indicated the possibility of foetal abnormalities such as spina bifida or Duchenne muscular dystrophy.

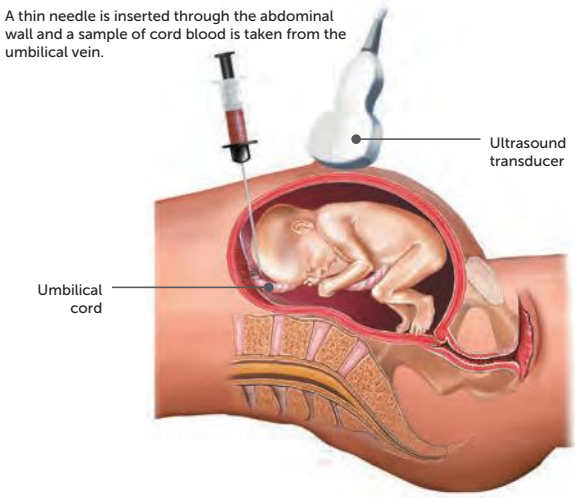
Foetal blood sampling

Foetal blood may be sampled to:

- diagnose chromosomal abnormalities
- diagnose foetal anaemia
- check foetal oxygenation
- identify infections
- give medications.

The blood may be taken from:

- the umbilical cord via percutaneous umbilical cord blood sampling (PUBS), where a needle is inserted through the abdominal wall and uterus into the umbilical vein
- a foetal blood vessel, usually the liver or heart, via a fetoscope.



Visuals Unlimited/Nucleus Medical Art

FIGURE 14.15
In percutaneous umbilical cord blood sampling, a sample of blood is taken from the umbilical vein of the foetus

The results from foetal blood sampling are obtained within a few days, as opposed to a few weeks for other forms of diagnostic testing such as amniocentesis. However, the risk of miscarriage is higher: 1–2%. There is also a risk of infection, blood loss and premature rupture of the amniotic sac.

Biochemical analysis

The assessment of marker proteins occurs with all newborns in Australia and in many other countries. This technique is used to detect phenylketonuria (PKU), either by testing the blood for excessive amounts of phenylalanine or by analysing the urine for phenylpyruvic acid. Another marker protein is alpha-fetoprotein (AFP), which can be measured in samples of amniotic fluid. The concentration of this protein is very high when the foetus has a malformation of the spinal cord, such as spina bifida.

DNA probes

DNA probes are a more recent innovation that enables the detection of a range of genetic disorders, such as Duchenne muscular dystrophy and thalassaemia. The probes are based on recombinant DNA technology. A segment of DNA is used that is structurally identical to the gene being tested. Some of the units in the DNA segment are 'labelled' with a dye or radioisotope. This DNA probe is then joined to the gene in question. If the gene is normal, the DNA probe joins with the DNA segments with which it is structurally identical and shows them up. If it is an abnormal gene, it does not show up and is identifiable as a gap in the DNA being tested.

Key concept

Advances in technology have led to the development of a range of monitoring and diagnostic techniques. The choice of method is dependent on the risks and necessity of the test.

Questions 14.2

RECALL KNOWLEDGE

1. Describe how an ultrasound image is produced.
2. List six reasons why a foetal ultrasound may be performed.
3. Describe how an amniocentesis is able to identify chromosomal, genetic or neutral abnormalities.
4. What does 'CVS' stand for?
5. State the reason for foetal monitoring.
6. Describe the use of a fibre-optic scope to study the foetus.
7. List the places where foetal blood is collected from for testing.
8. Identify the most common use of biochemical analysis for newborn babies in Australia.

APPLY KNOWLEDGE

9. Use a table to compare and contrast the uses, advantages and disadvantages of the different techniques used to monitor and diagnose foetal health.
10. Explain why an ultrasound is often used during an amniocentesis.
11. An amniocentesis is often performed between 16 and 20 weeks of gestation. Suggest why it is not performed at other times.
12. Explain why chorionic villus sampling may be preferred over amniocentesis, despite the higher risk of miscarriage.
13. A mother's blood may be tested to screen for some disorders. Justify the importance of this test, and discuss its advantage over foetal blood sampling.

WS

14.2 Foetal health

9780170449090

CHAPTER 14 ACTIVITY

ACTIVITY 14.1 Should we use assisted reproductive technologies?

In society, there are many different viewpoints regarding the use of assisted reproductive technologies.

What to do

1. Allocate one of the following roles to each member of the class. Ideally, there should be more than one person for each role.
   - A childless couple who have been trying to start a family for several years
   - A female who is about to undergo chemotherapy due to breast cancer
   - A person who was born because of the use of a reproductive technology
   - A doctor specialising in reproductive technology
   - A member of the public opposed to the use of reproductive technologies due to the fate of excess embryos
   - A single female who would like to have a child
   - A member of the clergy opposed to artificial insemination by donor and IVF
   - A scientist researching the improvement of reproductive technologies
   - A member of the public worried about the morality of choosing the sex of a baby
   - A person concerned about the poor success rate of IVF and the high financial cost
   - A same sex couple who would like to have a child
   - A couple who have not been able to have a child, despite trying for a number of years, but who are practising Catholics
2. Work with the other students who have the same role to brainstorm the viewpoints of the person/couple. You may need to conduct research so that your viewpoints are accurate regarding scientific, ethical, moral, religious and economic issues.
3. Hold a class discussion on the issues involved in the use of assisted reproductive technologies in humans. Contribute to the discussion from the viewpoint of your role, not your own personal viewpoint. You could consider questions such as:
   - Why do people's opinions differ about what should be permitted using reproductive technologies?
   - How can society best consider the wide range of views that people hold on these issues?
   - Who should be allowed to decide whether reproductive technologies are used?
   - What are the responsibilities of the scientists who research and develop reproductive technologies?
   - Who should set standards for laboratories and doctors involved in using modern reproductive technologies?
   - Who has the right to decide whether a particular person or couple should be allowed to use a particular reproductive technology?
   - Because of the high cost involved, should a limit be applied to the number of times a couple can use a particular procedure?
4. After listening to the opinions expressed during the discussions, prepare a list of arguments for and against the use of reproductive technologies in humans.

9780170449090

CHAPTER 14 SUMMARY

- Infertility is the inability to conceive despite frequent unprotected sexual intercourse.
- Anything that reduces the chance of the sperm reaching and fertilising the egg, or the fertilised egg implanting, will affect fertility.
- To fertilise an egg, sperm need to be in sufficient numbers, move forward and penetrate the layers of the egg.
- Blockages of the tubes, hormonal imbalances, antibodies affecting the sperm and low semen flow can affect male fertility.
- The number of healthy eggs in females decreases with age. Once menopause has been reached, ovulation stops.
- Polycystic ovarian syndrome results in many follicles forming, but failing to mature and release eggs.
- Endometriosis may block the uterine tubes, while some fibroids distort the uterus and uterine tubes.
- Problems with the menstrual cycle affect fertility by changing the development of the endometrium.
- Infertility treatments vary depending on the cause of the problem.
- Surgery may be used to correct structural problems and to collect sperm in males unable to ejaculate sufficient sperm.
- Ovulation tracking allows sexual intercourse to occur at a time when fertilisation is most likely.
- Medication can induce ovulation so that the egg is able to meet the sperm.
- Artificial insemination makes it more likely that the sperm will meet the egg by depositing it in the uterus.
- Assisted fertilisation techniques help fertilisation to occur.
- Gamete intrafallopian transfer involves the sperm and egg being collected and analysed before being mixed and deposited in the uterine tubes.
- In vitro fertilisation involves the sperm and the egg being mixed and incubated to allow fertilisation to occur. Embryos can then be transferred into the uterus.
- Intracytoplasmic sperm injection involves the sperm being injected into the egg, and the resulting embryo being transferred into the uterus.
- Donor eggs, sperm or embryos may be used for couples who have problems with these structures. Surrogacy can be used for females who are unable to conceive or carry a baby.
- Factors to consider with fertility treatments include the fate of unused embryos, religious beliefs and cost.
- Technologies are used to monitor the health of a foetus during pregnancy.
- Ultrasounds use sound waves to gain a visual image of the foetus. This allows doctors to confirm the pregnancy and monitor the foetus's development.
- Amniocentesis, chorionic villus sampling and blood tests are used to collect samples for genetic analysis.
- Amniocentesis uses a needle to extract some of the amniotic fluid, which contains foetal cells.
- Chorionic villus sampling uses a needle to extract cells from the chorion.
- Some of the baby's DNA passes into the mother's blood. A blood test can be used to screen for some disorders. Abnormalities detected can be diagnosed with amniocentesis or chorionic villus sampling.
- Foetal monitoring records the heart rate. It is done with an electrocardiograph.
- Fetoscopes can be a stethoscope, used to listen to the heart rate; or a fibre-optic scope inserted into the uterus to look at the foetus.
- Foetal blood tests provide quick results but have greater risks than other tests. Blood can be taken from the umbilical cord, umbilical vein or a foetal blood vessel.
- Testing blood or amniotic fluid can identify the presence of proteins indicating diseases such as phenylketonuria or spina bifida.
- DNA testing is able to detect disorders such as Duchenne muscular dystrophy and thalassaemia.

9780170449090

CHAPTER 14 GLOSSARY

Amniocentesis A technique in which a small amount of the amniotic fluid surrounding a foetus is removed and examined for indications of possible defects in the foetus

Artificial insemination Introducing semen from a donor into the vagina using a syringe, to bring about fertilisation

Assisted reproductive technology (ART) Methods developed to assist couples to have a child, including in vitro fertilisation and intrafallopian transfer

Chorionic villus sampling (CVS) A technique in which foetal cells are removed from the chorion and examined for indications of possible defects in the foetus

DNA probe A section of DNA that complements the gene of interest

Electrocardiogram (ECG) A record of the electrical activity of the heart

Electrocardiography The process of obtaining an electrocardiogram

Endometriosis A condition where tissue similar to the lining of the uterus grows outside the uterus

Fetoscope The instrument used to examine a foetus in utero

Fetoscopy The direct visual examination of a foetus through a small telescope-like instrument

Fibroids A non-cancerous growth around the uterus

Gamete intrafallopian transfer (GIFT) A method of assisted reproductive technology; a variation of IVF where sperm and eggs are mixed immediately after collecting the eggs

Infertility Not being able to get pregnant despite having frequent, unprotected sex

Intracytoplasmic sperm injection (ICSI) A procedure whereby a single sperm is inserted into a single egg

Intrauterine insemination (IUI) Placing the sperm inside the female's uterus

In vitro fertilisation (IVF) Fertilisation outside the body of the female

Polycystic ovarian syndrome (PCOS) A hormonal condition that causes changes to the menstrual cycle as well as cysts on the ovaries

Surrogacy When a woman bears a child for someone else

Ultrasound A technique in which inaudible, high-frequency sound waves are used to produce an image, especially of the foetus during pregnancy

9780170449090

CHAPTER 14 REVIEW QUESTIONS

Recall

1 List factors that may lead to infertility.
2 State what 'GIFT', 'ICSI', 'IVF' and 'IUI' stand for.
3 Describe the steps that occur during an IVF cycle.
4 What is foetal monitoring, and why is it used?
5 What alternatives are there for the unused embryos from IVF?
6 In what situations would a doctor advise a female patient to undergo genetic screening or counselling? What testing procedures could the doctor suggest?
7 Describe the procedure used for intrauterine insemination.

Explain

8 Explain why sperm factors account for approximately 40% of infertility cases.
9 Explain the procedure used in in vitro fertilisation.
10 Explain why ICSI is recommended for couples where poor sperm motility affects the possibility of conception.
11 Explain why surrogacy may be the most suitable option for a person or couple wanting to have a child.
12 Explain the conditions where donor sperm would be recommended.

Apply

13 List the considerations that would be factored into a decision regarding the use of assisted reproductive technologies.
14 Use a table to compare and contrast the methods used for foetal monitoring.
15 Explain why amniocentesis is offered to mothers aged 37 or over but is not routinely offered to mothers in their twenties.
16 If a child is born to a surrogate mother, will that child show any resemblance to the surrogate mother? Give reasons for your answer.

Extend

17 If it only takes one sperm to fertilise an egg, why is a sperm count of 15 million per millilitre considered a low count?
18 Discuss why it is important that couples who donate embryos are able to stipulate some characteristics of the female getting the embryo. Suggest some characteristics that may be factors.
19 New ethical and legal issues are arising with the more widespread use of reproductive technologies. In the United States, fertility clinics sell eggs and sperm from donors with specific attributes. They also advertise for donors with particular characteristics, such as being tall, with an athletic build and no major family medical problems. Consider the ethical and legal problems that shopping for gametes might bring. For example, are gametes to be considered like any other commodity? And if a couple have paid for gametes to produce a bright and athletic child, what legal recourse should they have if the child does not meet their expectations? List all the ethical and legal issues you think could arise from the advertising of, or for, gametes. Compare your list with those of other members of your class. You may wish to debate the issues involved.
20 Embryos resulting from IVF are tested genetically before they are implanted into the mother's uterus. An embryo

9780170449090

found to have a genetic disorder would probably not be used for implantation. What are some of the moral and ethical issues associated with disposing of unwanted embryos?

21 Amniocentesis and chorionic villus sampling are both invasive diagnostic tests that have complications associated with them that may cause harm to the foetus. A cervical-screening-type test is being developed to diagnose genetic abnormalities. Find out the progress being made on developing such a test and how long it is likely to be before it comes into general use. Are other non-invasive techniques being developed?

9780170449090

15 GENETICS CAN BE USED TO UNDERSTAND THE TRAITS OF INDIVIDUALS AND FAMILIES

UNIT 2 CONTENT

SCIENCE INQUIRY SKILLS

- conduct investigations, safely, competently and methodically for the collection of valid and reliable data
- represent data in meaningful and useful ways; organise and analyse data to identify trends, patterns and relationships; qualitatively describe sources of measurement error and uncertainty and limitations in data; and select, synthesise and use evidence to make and justify conclusions
- select, construct and use appropriate representations, including models of DNA replication, transcription and translation, Punnett squares, pedigrees and karyotypes, to communicate conceptual understanding, solve problems and make predictions

SCIENCE UNDERSTANDING

Cell reproduction

- variations in the genotypes of offspring, including gender, arise as a result of the processes of meiosis and fertilisation

Types of inheritance

- probable frequencies of genotype and phenotype of offspring can be predicted using Punnett squares and by taking into consideration patterns of inheritance, including the effects of dominance, co-dominance, autosomal or sex-linked alleles, and multiple alleles: Huntington's disease, phenylketonuria (PKU), ABO blood groups, red–green colour blindness/haemophilia show different inheritance patterns
- pedigree charts can be constructed for families with a particular genetic disorder and can be used to reveal patterns of inheritance and assist in determining the probability of inheriting the condition in future generations
- DNA profiling identifies the unique genetic make-up of individuals and can be used in determining parentage

Source: School Curriculum and Standards Authority, Government of Western Australia

9780170449090

In any population, it is easy to see that the individuals differ from one another. In Chapters 10 and 11, you learnt about meiosis, and how it results in each gamete containing only one chromosome from each homologous pair. You also learnt about how the processes of crossing over, non-disjunction and random assortment of chromosomes result in variation between gametes. In Chapter 12, you learnt about the random way in which sperm fertilises an egg. However, when you look at individuals in the same family it is obvious that there is a reason for the similarities between them. In this chapter, we will look at understanding the variations between related individuals.

Newpix/Scott Fletcher

FIGURE 15.1 Five generations of one family. Family characteristics are passed from one generation to the next through inheritance

15.1 MENDELIAN INHERITANCE

From very early times, farmers realised that many characteristics of domesticated plants and animals were passed from parent to offspring. Exactly how these characteristics, or **traits**, were transmitted was not clear. It was thought by many that offspring were simply a blend of the characteristics of the two parents. However, this was not the case. The first clear explanation of patterns of inheritance was provided by an Austrian monk, Gregor Mendel, in 1865. After spending two years at the University of Vienna, he returned to his monastery as a schoolteacher. Here he was able to link his two loves, nature and mathematics, through a careful study of the reproductive behaviour of pea plants. After 10 years of research, Mendel put forward two principles relating to inheritance:

1. The various hereditary characteristics were controlled by factors (now called **genes**) that occurred in pairs.
2. During the formation of the gametes (in humans, the eggs and the sperm), the pairs of factors separate. Each gamete receives only one set of factors, or genes, with the other set going to another gamete. Gametes unite at fertilisation, allowing different combinations of genes to come together.

Getty Images/Bettmann

FIGURE 15.2 Gregor Mendel

Gregor Mendel
This website features more information on the life of Gregor Mendel.

Mendel's findings went unnoticed for 35 years before their significance was fully appreciated. At the same time as his work was being rediscovered, scientists were making considerable advances in cytology, the study of cells. A young American graduate student, Walter Sutton, was able to link the work of Mendel to that of the cytologists. His observations of the behaviour of chromosomes during meiosis, and Mendel's speculation on the separation of the hereditary factors during the formation of gametes, led Sutton to suggest that the hereditary factors, or genes, were located in the chromosomes. This important hypothesis, contained in a research paper he published in 1903, led to the chromosome theory of heredity.

Mendel's experiments

Mendel conducted breeding experiments with the edible garden pea, *Pisum sativum*, and was impressed by the fact that it possessed a number of characteristics, or traits, that were expressed in contrasting forms. He studied seven pairs of contrasting characteristics in which the alternatives were easily identifiable.

FIGURE 15.3
The seven pairs of contrasting characteristics in garden peas studied by Mendel

CHARACTERISTIC STUDIED	DOMINANT CHARACTER	RECESSIVE CHARACTER
Seed shape	Round 	Wrinkled 
Seed colour	Yellow 	Green 
Seed-coat colour	Coloured 	White 
Pod shape	Inflated 	Constricted 
Pod colour	Green 	Yellow 
Flower position	Axial 	Terminal 
Stem length	Long 	Short 

Before beginning an experiment, Mendel made sure his plants were **pure-breeding** for the characteristic he wished to study. Pure-breeding plants are those that produce the same characteristic in each succeeding generation when bred among themselves. He then crossed, or interbred, plants with contrasting traits. For example, plants pure-breeding for yellow-coloured seeds were crossed with plants pure-breeding for green seeds. He found that the offspring (or **progeny**) resembled only one of the parents. In the example given, the offspring were all plants that produced yellow seeds. These offspring are referred to as **hybrids** because they have genetic information for green seed colour as well as genetic information for yellow seed colour, even though they are all yellow. Thus, only *one* of the pair of contrasting characteristics appeared in the offspring. Mendel referred to the characteristic shown by the hybrid as the **dominant trait** because it masked the appearance of the other characteristic, which he called the **recessive trait**.

9780170449090

When Mendel allowed the hybrid plants to self-pollinate, a second generation of plants was produced. In this generation the characteristics reappeared in the ratio of about three with the dominant trait for every one with the recessive trait (3:1). From these results, Mendel concluded that the hereditary factors, or genes, were unchanged as they passed from one generation to the next. He further reasoned that each pea plant had two hereditary factors for each characteristic under study. During the formation of gametes, these factors are separated (or, as Mendel called it, segregated), with each gamete receiving only one factor, or gene, for each trait. This is known as the **principle of segregation**. As offspring are formed by the union of a male and a female gamete, each offspring receives one gene for each characteristic from each parent.

Mendel and his peas
This website has more information about Mendel and his study of peas.

Key concept

The principle of segregation means that, at fertilisation, an offspring receives a gene for each trait from each parent.

Genotypes and phenotypes

A cross is the mating of two organisms. In a **monohybrid cross**, only one pair of contrasting characteristics is studied – for example, yellow and green pod colour in peas, or tongue rolling and non-rolling in humans.

It is much easier to refer to the genes by a letter than by name. Therefore, the genes for a particular characteristic are represented by two letters, one for the gene that originated from the female parent and one for the gene that originated from the male parent. If the gene is a dominant one, it is shown as a capital letter; if it is a recessive one, a lower-case letter is used. For example, in the garden pea, green pod colour is dominant to yellow, so the gene for green pod colour is represented with a capital *G* and the recessive gene for yellow pod colour with a lower-case *g*.

For pure-breeding plants of green pod colour, the symbols used are *GG*; pure-breeding plants of yellow pod colour are *gg*. Hybrids, with one of each gene type, have the symbols *Gg*. The alternative forms of the gene for pod colour – in this case, *G* and *g* – are called **alleles**. The combination of alleles for a particular trait is called the **genotype**. For a pair of contrasting characteristics, the two alleles may occur in one of three possible combinations: *GG*, *gg* or *Gg*. In two of these three genotypes the alleles are the same (*GG* and *gg*). These are described as **homozygous**, whereas the hybrid, *Gg*, with one of each allele, is termed **heterozygous**. The three genotypes listed produce only two types of pod colour in garden peas – green for *GG* and *Gg*, and yellow for *gg*. This physical appearance, or what the pods look like, is called the **phenotype**. These and other terms used to describe inheritance are defined in Table 15.1.

Key concept

The phenotype is the physical appearance of a trait as determined by the genotype, the combination of alleles. If the alleles are the same, the genotype is homozygous; if the alleles are different, the genotype is heterozygous.

TABLE 15.1 Terms relating to Mendelian genetics

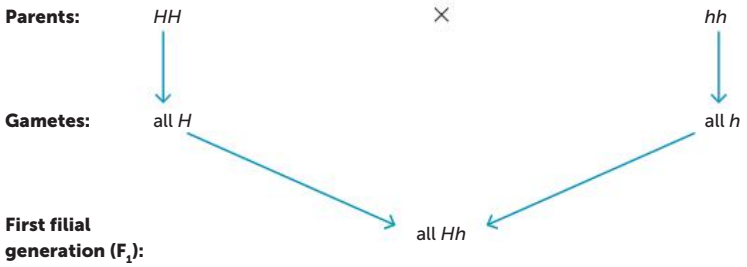
TERM	MEANING
Gene	The factor that determines an inherited characteristic; located in the chromosomes
Allele	An alternative form of a gene (e.g. the gene for pod colour in peas has two alleles, green and yellow)
Dominant	An allele that masks the effect of another allele (e.g. a pea plant with alleles for green and yellow pods will produce green pods because green is dominant to yellow)
Recessive	An allele that is masked by the alternative, dominant allele (e.g. the allele for yellow pod colour is masked by the allele for green pod colour)
Homozygous	The situation where an individual has the same alleles for a particular characteristic; also called pure-breeding (e.g. a pea plant with two alleles for green pod colour is homozygous green)

TABLE 15.1 (*Continued*)

TERM	MEANING
Heterozygous	The situation where an individual possesses different alleles for a particular characteristic; also called hybrid (e.g. a pea plant with alleles for both green- and yellow-coloured pods is heterozygous)
Phenotype	The physical appearance of an individual as determined by the expression of the alleles for that characteristic (e.g. a pea plant with alleles for green-coloured and for yellow-coloured pods will have the phenotype green pods)
Genotype	The genetic make-up of an individual as determined by the alleles for the characteristic being considered (e.g. a pea plant with one allele for green-coloured pods and one for yellow-coloured pods will have the heterozygous genotype)

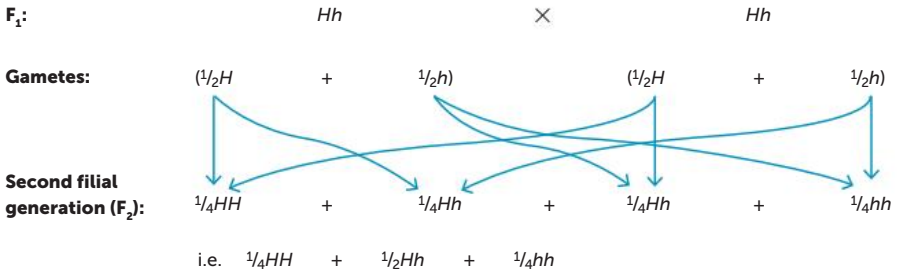
Using these terms, we can now look at some examples. Another of Mendel's crosses involved crossing pure-breeding long-stemmed pea plants with pure-breeding short-stemmed plants. The homozygous long-stemmed plants can be represented by the letters *HH*; the homozygous short-stemmed plants by *hh*. During the formation of gametes by meiosis, the pairs of chromosomes separate, with one of each pair going to each gamete. This means that the pairs of alleles segregate with only one allele for a characteristic carried by each gamete. In this case, all the gametes of the long-stemmed plants will have the allele *H*, and all the gametes of the short-stemmed plants will have the allele *h*. Therefore, the offspring will all be the same, *Hh*. As the allele for long stem is dominant, the offspring will all appear as long-stemmed plants. The allele for short stem is masked, as it is a recessive characteristic. The offspring are referred to as the **first filial generation**, denoted by the symbol F_1. This cross is shown diagrammatically in Figure 15.4.

FIGURE 15.4 First filial generation



If the first filial generation is self-pollinated, a second set of offspring is produced. This is the **second filial generation**, or F_2. In this case, the hybrid F_1 plants produce gametes with half containing the allele for long stem and half containing the allele for short stem. Every male gamete has an equal chance of meeting a female gamete, so that the chance of getting particular genotypes in the second filial generation can be shown mathematically, as in Figure 15.5.

FIGURE 15.5 Second filial generation



From the final equation in Figure 15.5 you will notice that one-quarter of the F_2 are homozygous for long stems (*HH*), one-half are heterozygous (*Hh*), and one-quarter are homozygous for short stems (*hh*). However, the homozygous long-stemmed plants and the heterozygous plants would appear the same; that is, three-quarters of the second generation would appear long-stemmed and one-quarter short-stemmed.

Activity 15.1
Investigating Mendelian genetic principles in Martians

Questions 15.1

RECALL KNOWLEDGE

1 What is another term for trait?
2 What organisms were used in Mendel's experiments?
3 Describe the difference between pure-breeding and hybrid.
4 Define:
   a dominant trait
   b recessive trait.
5 Describe the principle of segregation.
6 Describe the difference between:
   a homozygous and heterozygous
   b genotype and phenotype.

APPLY KNOWLEDGE

7 Explain why it was important that Mendel's work matched the observations of Sutton's work on cells.
8 Explain why it was important that the pea plants originally used by Mendel were pure-bred.
9 Explain why the letter x is not a suitable choice to represent an allele.
10 Classify each of the following genotypes as homozygous or heterozygous:
   a Rr
   b GG
   c Tt
   d aa
   e EE.
11 State whether each of the following is a genotype or phenotype.
   a curly hair
   b *Rr*
   c *HH*
   d blue eyes.
12 In Mendel's experiments, there were no short plants in the first filial generation. However, approximately one-quarter of the plants in the second filial generation were short. Explain this observation.

15.2 MODELLING INHERITANCE

It is useful to construct models to better understand and represent concepts, and inheritance is no exception. In genetics, we can use Punnett squares to model an individual cross and a pedigree chart to show the appearance of a certain trait in a family.

Punnett squares

A **Punnett square** can be used to model a cross and, therefore, to calculate the probability of genotypes and phenotypes of offspring. The Punnett square is named after R. C. Punnett, a British geneticist of the early 20th century, who devised the square to use in his work on heredity.

In a Punnett square for a monohybrid cross:

- Draw a square with two columns and two rows.
- Write the alleles for one parent above the columns – one allele above each column.
- Write the alleles for the other parent to the left of the rows – one allele for each row.
- For each box, write the combination of alleles for that row and column. Each box represents a possible genotype for the offspring.
- Calculate the probability of each genotype and, hence, phenotype.

9780170449090

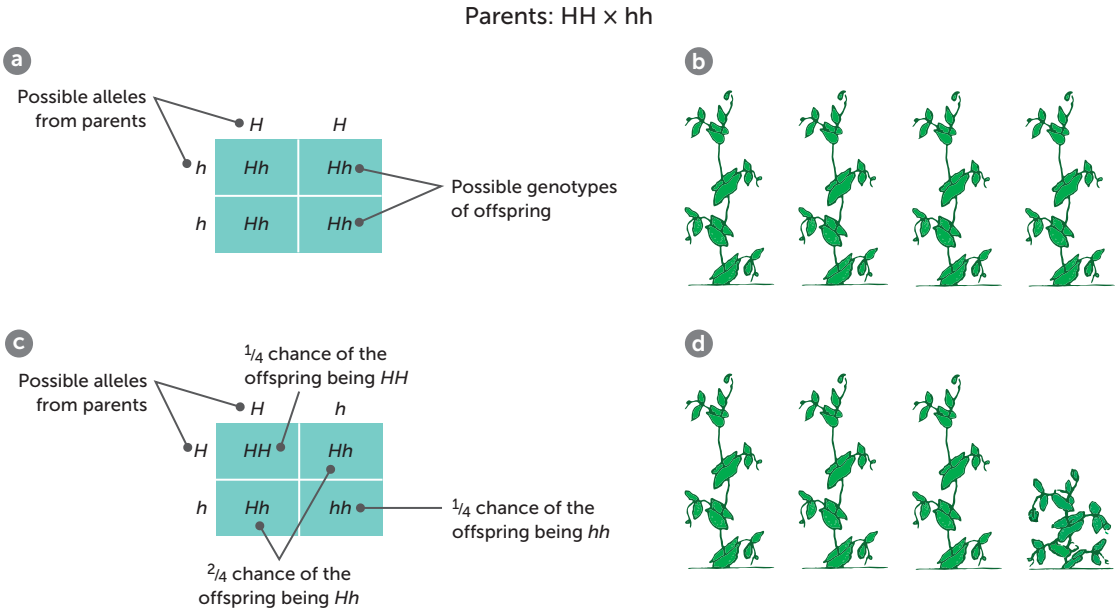


FIGURE 15.6 a Punnett square for a cross between parents with *HH* and *hh* genotypes. There is a 100% chance of the offspring being *Hh;* **b** F_1 genotype are all *Hh* and, therefore, long-stemmed plants; **c** Punnett square for the F_2 generation – a cross between *Hh* and *Hh*. The probabilities of genotypes for the offspring are ¼ *HH*, ²/₄ *Hh* and ¼ *hh*; **d** F_2 phenotypes are ³/₄ long-stemmed plants (due to ¼ *HH* and ²/₄ *Hh*) and ¼ short-stemmed plants (due to ¼ *hh*)

Each pea plant produces a great many seeds and, therefore, many potential offspring, so it is appropriate to talk about the proportion of each genotype or phenotype that should occur in the offspring of a cross. However, it is more accurate to refer to the probability of an offspring having a certain genotype of phenotype. This means that the chance that any one seed will grow into a long-stemmed plant is ¾, or 0.75, or 75%, while the probability that a seed will produce a plant that is long-stemmed and homozygous for the long-stem allele is ¼ (0.25 or 25%).

The principles of Mendelian inheritance and Punnett squares can also be applied to human traits. While some characteristics are influenced by more than one gene, and are beyond the scope of this course, there are many examples that are appropriate. For example, long eyelashes are dominant over short eyelashes. If a mother with long eyelashes who is known to be heterozygous and a father with short eyelashes are expecting a child, we can use a Punnett square to predict the possible eyelash length of the offspring.

	e	*e*
E	*Ee*	*Ee*
e	*ee*	*ee*

FIGURE 15.7 Punnett square for a cross between *ee* and *Ee*. The F_1 have a 50% chance of having long eyelashes and a 50% chance of having short eyelashes

Key concept

Punnett squares are used to predict the probability of possible genotypes and phenotypes for offspring of certain parents.

9780170449090

TABLE 15.2 Some inherited traits in humans

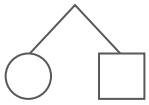
DOMINANT	RECESSIVE
Free earlobes	Attached earlobes
Broad lips	Thin lips
Long eyelashes	Short eyelashes
Broad nostrils	Narrow nostrils
Abundant body hair	Little body hair
Curly hair	Straight hair
Mongolian eye fold	No eye fold
Astigmatism	Normal vision
Roman nose	Straight nose
Huntington's disease	No disorder
Achondroplasia	Normal build
Normal enzyme production	Phenylketonuria
Normal pigmentation	Albinism
Tongue rolling	Non-rolling

Pedigrees

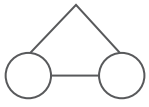
A Punnett square is an effective way to model a single possible cross; however, it is not useful to look at the phenotypes that have been produced in a family. A **pedigree**, or family tree, is used for this purpose.

A number of conventional symbols are used in the construction of such pedigrees.

- Males are represented by squares (□).
- Females are represented by circles (○).
- Those with the particular characteristic under study are shaded (■ or ●).
- A union (or marriage) is represented by a horizontal line joining the symbols (□—○).
- A union between two close relatives, usually cousins, called a **consanguineous union**, is shown by double horizontal lines (□=○).
- A vertical line extending down from the horizontal one connects to the children produced by that union.
- If there are a number of children from a particular union, each is connected by a vertical line to a horizontal one, which in turn links up with the line joining the two parents. The children are shown in order of birth, from left to right.
- Twins have lines coming from the same point on the horizontal line.



- Identical twins have a horizontal line joining them.



- A death is indicated by a diagonal line drawn through the symbol.

- The generations are numbered with Roman numerals, which are written on the left-hand side.
- Individuals within a generation are numbered from left to right (starting with 1 for each generation).

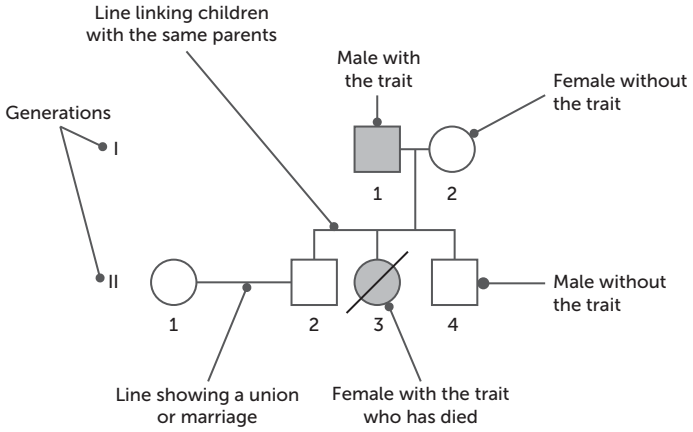


FIGURE 15.8 A pedigree for two generations of a family

Key concept

Pedigrees are used to investigate the patterns of inheritance of traits over generations of a family.

One human characteristic that is obvious to any observer is skin colour. This trait depends mainly on the amount of melanin, a yellow-black pigment produced by special skin cells called melanocytes. The production of melanin by the melanocytes is dependent on particular enzymes. In a very small number of cases, humans fail to synthesise one of the enzymes and pigmentation does not occur. These individuals, who are called **albinos**, have white skin, white hair and pink eyes (due to the reflection from blood vessels in the eye).

Production of melanin is a good example of how simple dominance operates in humans. Two alleles are involved in pigmentation. The dominant allele controls normal enzyme production and thus the presence of melanin. The recessive allele causes abnormal enzyme production. Genetically, albinos are homozygous for the recessive allele and therefore do not produce any pigment.

Figure 15.9 illustrates four generations of a family in which one of the original parents was an albino. Albinism did not occur again in this family until the cousins in the third generation married and produced children.

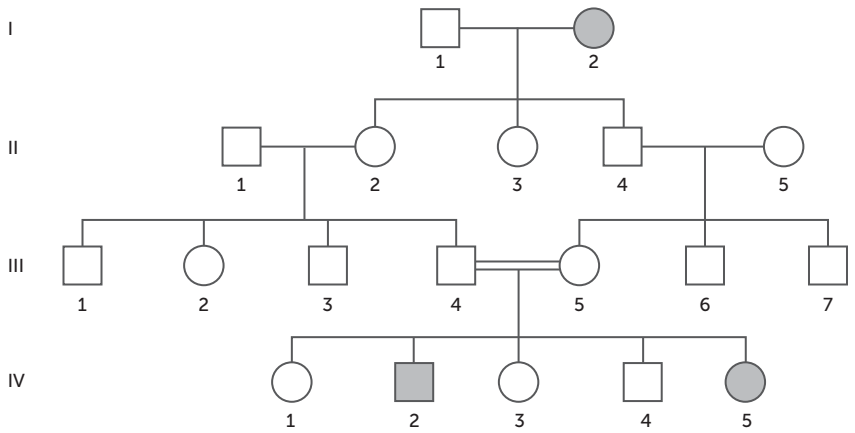


FIGURE 15.9 A pedigree with one original parent an albino

We can use this pedigree to work out the genotypes, with respect to skin pigmentation, of all the individuals in the family. We will represent the dominant allele for normal pigmentation by *A* and the recessive allele for albinism by *a*.

The Punnett squares in Figure 15.10 show all the possible combinations of parents.

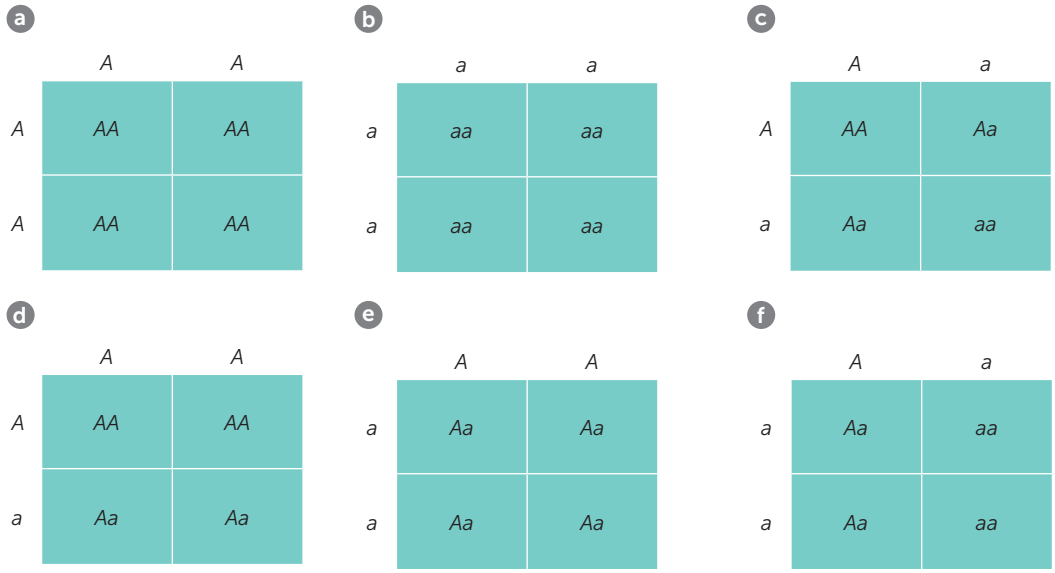


FIGURE 15.10
Possible combinations of parents:
a Parents *AA* and *AA*
b Parents *aa* and *aa*
c Parents *Aa* and *Aa*
d Parents *AA* and *Aa*
e Parents *AA* and *aa*
f Parents *Aa* and *aa*

We can use knowledge of whether the trait is dominant or recessive, the presence or absence of the trait along with the information from the Punnett squares to infer the genotypes of some or all of the family members. Recalling that albinism is a recessive trait, the pedigree tells us that:

- Individuals who are albinos must be homozygous recessive (*aa*).
- As the cousins in the third generation produced two children with albinism, the cousins must each have been heterozygous (*Aa*).
- It follows, therefore, that at least *one* of each of the cousin's parents (in generation II) must have been heterozygous as well, otherwise the cousins could not have inherited the recessive allele for albinism.
- Because the female in generation I is an albino, we know that the parents of the cousins (II2 and II4), who are brother and sister, must be heterozygous.

Figure 15.11 shows what we have worked out so far.

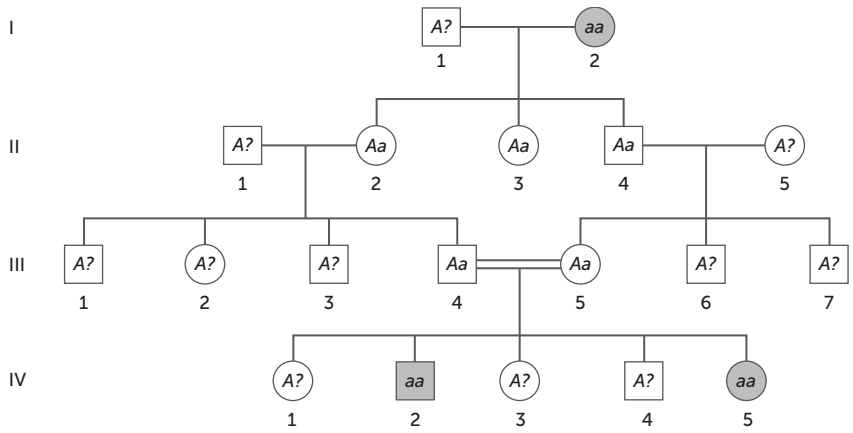


FIGURE 15.11
Genotypes for albinism

Without further information it is difficult to determine the genotypes of some of the individuals. We do not know if those marked *A*? are *AA* or *Aa*. More than likely, those marked as *A*? in the first and second generations were *AA*, otherwise albinos could have been expected in the third generation. However, we cannot be sure without further information.

Another human characteristic that can be traced through a family is whether the earlobes are attached or free. Generally speaking, those with free earlobes carry a dominant allele *F*, and such individuals may be of two genotypes: *FF* or *Ff*. Attached earlobes are a recessive trait and, therefore, have the genotype *ff*. The pedigree in Figure 15.13 shows three generations of a family. Those with free earlobes are shown as open symbols; those with attached earlobes are shaded.

9780170449090

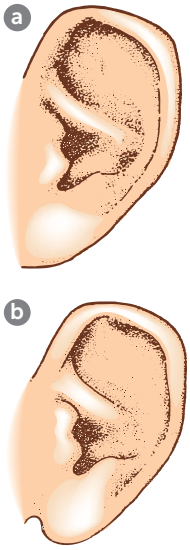


FIGURE 15.12
a Attached earlobe;
b Free earlobe

Once again, all the individuals shown shaded must be homozygous recessive (*ff*), whereas the parents in the second generation (II1, II2, II4 and II5) must all be heterozygous (*Ff*) as they have the dominant trait, but pass on a recessive allele. Apart from knowing that all other individuals have at least one *F* allele, it is difficult to determine their exact genotypes. At least one of the original parents must have been heterozygous, to produce children who were heterozygous; however, without more information, we do not know which parent it is. Figure 15.14 summarises what we are able to determine from the information we have.

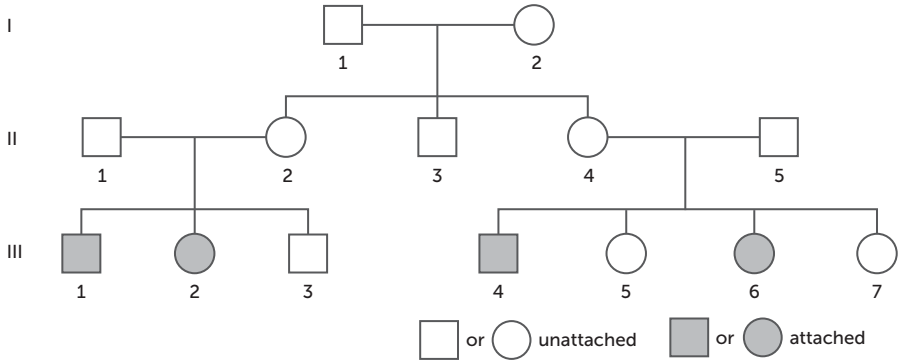


FIGURE 15.13 Pedigree for a family with attached earlobes appearing in the third generation

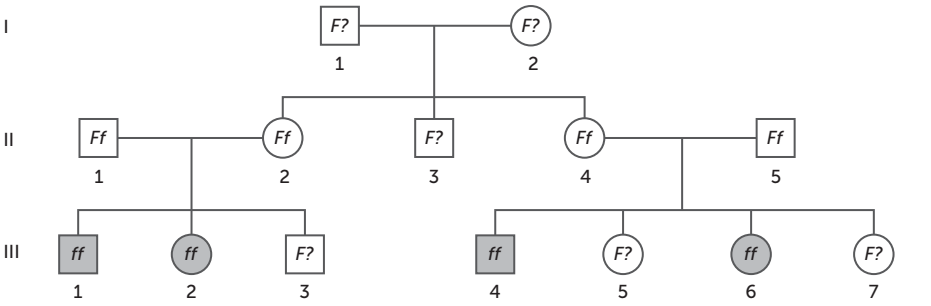
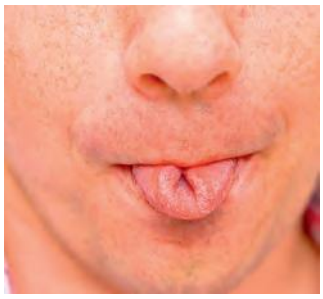


FIGURE 15.14 Genotypes for a family with attached earlobes appearing in the third generation

Tongue rolling in humans is a dominant characteristic. The ability to roll the tongue in three generations of a family was represented in the pedigree in Figure 15.16.



Shutterstock.com/Thoom

FIGURE 15.15 A tongue roller

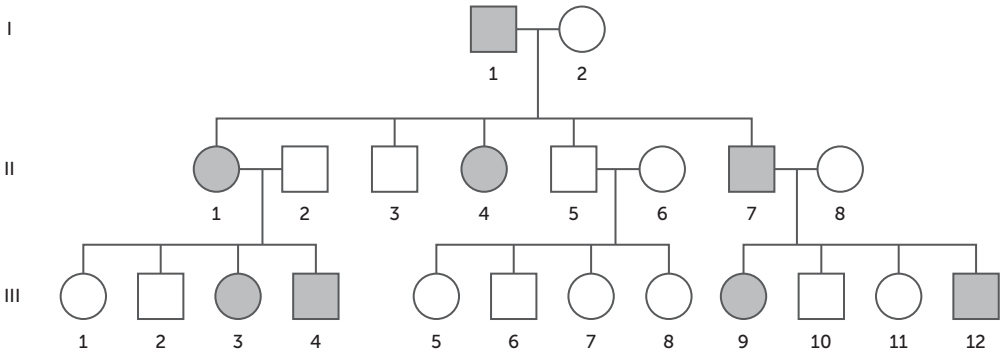


FIGURE 15.16 A pedigree for tongue rolling in three generations of a family

As the trait is dominant, the absence of it indicates that the individual is homozygous recessive. Any parents who are tongue rollers and have children who are not tongue rollers must be heterozygous in order to pass a recessive allele on to the child. And any tongue-rolling children with a parent who is not a tongue roller must also be heterozygous, as they could only receive a

recessive allele from that parent. With this information, we are able to determine the genotypes as shown in Figure 15.17.

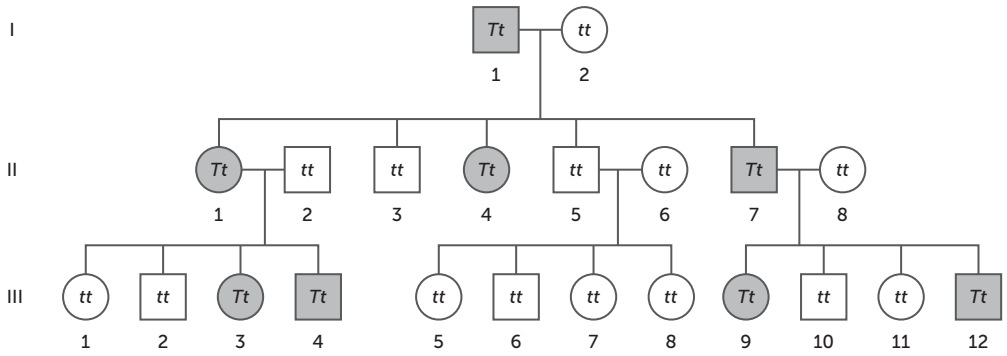


FIGURE 15.17 The pedigree for tongue rolling showing genotypes

Activity 15.2
Examining pedigrees

Questions 15.2

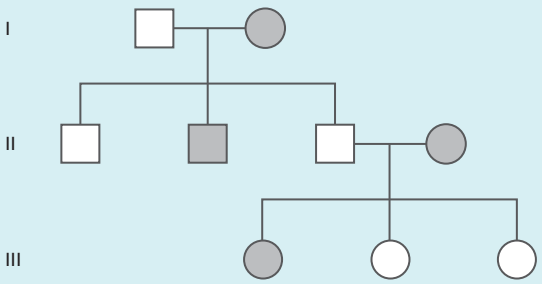
RECALL KNOWLEDGE

1. The probability of a child having a trait is 25%. What is this probability as a fraction?
2. How are each of the following represented in a pedigree?
   a. Male with the trait
   b. A deceased female with the trait
   c. Children from the same parents
   d. The generation
   e. A female without the trait
3. Define 'consanguineous'.

APPLY KNOWLEDGE

Use Table 15.2 to answer the following questions.

4. Suggest a genotype for someone with no eye folds.
5. Draw a Punnett square for each of the following crosses.
   a. $RR \times rr$
   b. $Mm \times Mm$
   c. $Ee \times ee$
6. What is the probability that a child will have astigmatism if neither of his parents have the condition? Explain your answer.
7. Two parents with broad lips have a child with thin lips. Use a Punnett square to determine the genotype of the parents.
8. Use the pedigree below to answer the following questions.



   a. How many females have the trait?
   b. If the trait is dominant, what is the genotype of the male in generation I?
   c. If the trait is recessive, what is the genotype of the:
      i. female in generation II?
      ii. unaffected females in generation III?

15.3 AUTOSOMAL INHERITANCE OF SINGLE-GENE DISORDERS

One of the most frequent reasons for constructing a pedigree for a family is to investigate the pattern of inheritance of a genetic disorder. **Single-gene disorders** are disorders caused by the inheritance of a single defective gene. The pattern of inheritance of single-gene disorders follows the basic laws of heredity already described. However, the severity of the disorder is often variable and difficult to predict. More than 4000 different disorders of this type have been identified in humans.

Humans have 46 chromosomes in all cells except gametes. These are composed of 23 pairs; 22 pairs are autosomal chromosome and 1 pair are sex chromosomes. Autosomal inheritance relates to traits, including disorders, that are inherited on the autosomal chromosomes.

Dominant, autosomal inheritance of single-gene disorders

Typically, traits that are controlled by dominant alleles are easily passed on, as only one allele is needed for the trait to be shown. However, dominant alleles that cause severe defects in people are rarely passed on, because people with such alleles frequently die before they have the opportunity to reproduce.

The following are some examples of genetic disorders caused by dominant alleles on the autosomal chromosomes:

- *Achondroplasia* – a form of dwarfism characterised by short limbs, a prominent head, normal intelligence and difficulty walking.
- *Facioscapulohumeral muscular dystrophy* – a rare form of muscular dystrophy affecting the facial muscles. Other muscles are gradually affected, making it difficult to raise the arms above the shoulders, to lift objects or to walk normally.
- *Huntington's disease* – an inherited disorder that results in the death of brain cells, causing changes to mood and mental ability as well as uncoordinated movement.
- *Neurofibromatosis* – affected individuals exhibit numerous tumours along the peripheral nerves. The tumours are composed of a dense proliferation of nervous and fibrous tissue, and cause abnormalities of the skin and flesh as well as distortions of bone structure.

Huntington's disease

Huntington's disease (formerly 'Huntington's chorea') affects between 5 and 10 people per 100 000 in developed countries, with symptoms usually appearing after 40 years of age. It is characterised by occasional involuntary flailing movements of the arms and legs. In addition, the person often has difficulty making voluntary movements of the limbs. Other symptoms include writhing movements of the hands, head, trunk and feet, and a progressive loss of the ability to think clearly, referred to as **dementia**.

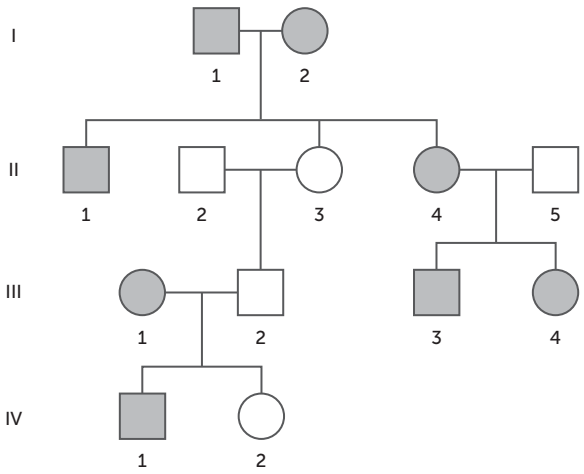


FIGURE 15.18 A pedigree of four generations of a family with a history of Huntington's disease

As Huntington's disease is controlled by a dominant allele, the condition is very likely to be passed on from one generation to the next. Because the condition does not become apparent until later in life, the children of a person with the condition may not be aware that they themselves may have inherited the disorder until after they have had children of their own.

9780170449090

Genetic screening is now available for people with a parent who either has Huntington's disease or died from it. Blood from participants and their close relatives is collected and analysed for the presence of three established gene markers. These tests establish an individual's risk of developing the condition. Because the condition is incurable, nothing can be done to help people diagnosed as having the disease, but those people can then make decisions about whether or not to have children.

Activity 15.3
Studying a family with Huntington's disease

Key concept

Huntington's disease is a single-gene disorder caused by a dominant allele. Symptoms of the disease are often not present until after the individual has had children, and so the dominant allele may be passed without the parents being aware of the possibility.

Recessive, autosomal inheritance of single-gene disorders

A person who has a heterozygous genotype has a recessive allele but does not show the recessive phenotype. They are known as a **carrier** for that characteristic. If both parents are carriers, there is a 25% chance that a child receives one defective allele from each parent. This results in the child being homozygous for the recessive allele and, therefore, being affected by the condition. If *N* is the normal allele and *n* the allele that causes an abnormality, this can be shown as illustrated in the Punnett square below.

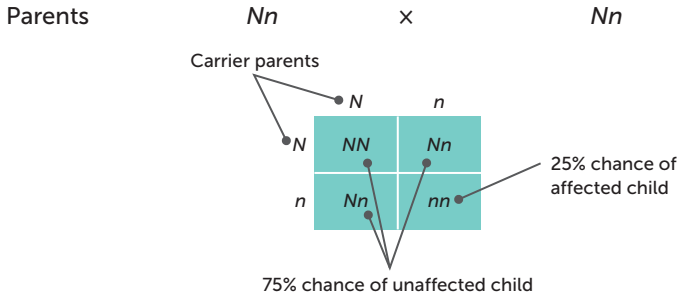


FIGURE 15.19 Two carrier parents have a 25% chance that a child will be affected by the recessive disorder

The incidence in the population of severe recessive disorders that are not linked to a sex chromosome is low, because it is unlikely that a carrier from one family will mate with another carrier of the same recessive condition. However, although a particular condition may be an inherited disorder, it could suddenly appear where there has been no previous family history of the disease. In a consanguineous marriage, because the couple are close relatives (e.g. cousins), the chance of them both being a carrier for a recessive allele is greater. Such marriages may occur for cultural or geographical reasons in certain populations. In these cases, there may be a higher incidence of a particular genetic disorder, as the related parents have received some of their genes from a common ancestor and so have a greater chance of being carriers of a gene for the same recessive condition. However, this is not often the case, and the chance of cousins marrying and having a child with a genetic disorder is little higher than if two strangers were to marry. Both Albert Einstein and Charles Darwin married their first cousins all their children were free of genetic disorders.

Phenylketonuria

Many recessive disorders produce serious abnormalities. **Phenylketonuria (PKU)**, which affects approximately 1 in 10 000 people in Australia, is a good example of a disease of this type. The gene concerned controls the production of an enzyme called phenylalanine hydroxylase. This enzyme converts the amino acid phenylalanine to tyrosine. If this enzyme is not present, then phenylalanine will accumulate in the bloodstream and become toxic. The toxicity results in damage to the growing brain, and thus produces extreme mental retardation, as well as a tendency towards epileptic seizures and a failure to produce normal skin pigmentation.

Phenylalanine is classed as an essential amino acid and so must be present in the diet. Enough of this amino acid must be present so that a child may grow. For those individuals born homozygous

recessive for PKU, excessive amounts of phenylalanine in the diet can be dangerous. Fortunately, the disease can be identified almost immediately after birth. A blood sample is usually taken by pricking the baby's heel (called a heel prick) within two to three days after birth. Special diets restricting the intake of phenylalanine and replacing it with substitutes can, if begun early in the child's life, largely, if not entirely, correct the symptoms.

Alamy Stock Photo/CROZSTUDIOS

FIGURE 15.20 A heel prick test is used to screen for a number of genetic disorders, including phenylketonuria

Cystic fibrosis

Cystic fibrosis is another disorder controlled by one recessive allele. Children with the condition suffer from chest infections, lack of digestive enzymes and increased salt loss. It is the most common lethal genetic disease in people of European origin. In Australia, 1 in 3700 people have cystic fibrosis and 1 in 25 are carriers. Once again, a blood sample is usually taken from the baby's heel within two to three days after birth. When a child is identified as having the disease, it is given a special diet low in fat and high in carbohydrate and protein. The diet is supplemented with pancreatic extract and large doses of vitamins A, D and K. This does not cure the disease, but it does enable the child to function as normally as possible.

Couples who are concerned that they may be carriers for cystic fibrosis can be tested via a laboratory test done on a sample of blood or saliva. If results show that both prospective parents are carriers, genetic counselling should be considered before starting a family.

Key concept

Phenylketonuria and cystic fibrosis are both recessive, single-gene disorders. For an individual to have the disorder, they must inherit a recessive allele from each parent.

Using pedigrees for single-gene disorders

You can use the criteria listed in Table 15.3 to make predictions about the chances of inheriting a single-gene disorder.

TABLE 15.3 Determining patterns of autosomal inheritance in pedigrees

DOMINANT TRAITS
Every person who shows a dominant characteristic need only have *one* allele for that characteristic, and every person possessing such an allele *must* show the characteristic.
A person with a dominant characteristic *must* have at least one parent with the characteristic. It *cannot* skip a generation.
Two people *with* a dominant characteristic *can* have a child *without* that characteristic.
Two people who do not possess a dominant characteristic *cannot* have a child with such a characteristic.
RECESSIVE TRAITS
Every person who shows a recessive characteristic *must* have two alleles for that characteristic.
A person with a recessive characteristic *does not* have to have a parent with the characteristic. It *can* skip a generation.
Two people *without* a recessive characteristic can have a child *with* the characteristic.
Two people *with* a recessive characteristic *cannot* have a child *without* the characteristic.

Activity 15.4
Investigating patterns of inheritance in heterozygous barley seeds

15.1 Pedigree charts

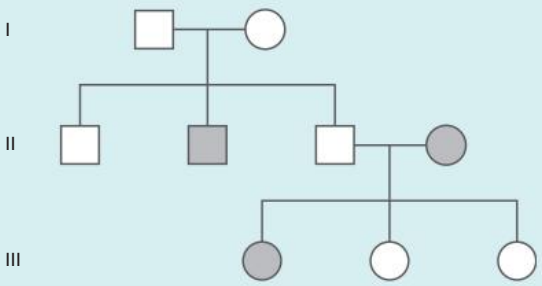
Questions 15.3

RECALL KNOWLEDGE

1. List five single-gene disorders.
2. Describe the symptoms of Huntington's disease and phenylketonuria.
3. Describe how someone can be a carrier of a genetic disorder.
4. Name the test used to screen for phenylketonuria.

APPLY KNOWLEDGE

5. Explain why disorders due to a recessive allele are passed on more frequently than those due to a dominant allele.
6. Discuss the use of genetic screening with regards to Huntington's disease. Include possible reasons people may choose, or not choose, to have screening done.
7. Explain why it is possible to be a carrier of a recessive disorder, but not a dominant one.
8. State whether the pedigree below is for a recessive or dominant disorder. Justify your answer.



15.4 SEX CHROMOSOMES

An examination of birth records for Australia this century would indicate that girls and boys are born in approximately equal numbers. However, a given family does not necessarily contain the same number of boys as girls. For centuries, people have tried to explain how the sex of a child is determined, and it was not until scientists began to examine the nuclei of cells that they realised the chromosome sets in the nuclei of cells of men and women were slightly different. In women, the 46 chromosomes in the

nucleus of each cell were in 23 matched pairs, whereas in men the 46 chromosomes were in only 22 matched pairs, the 23rd pair consisting of two unmatched chromosomes.

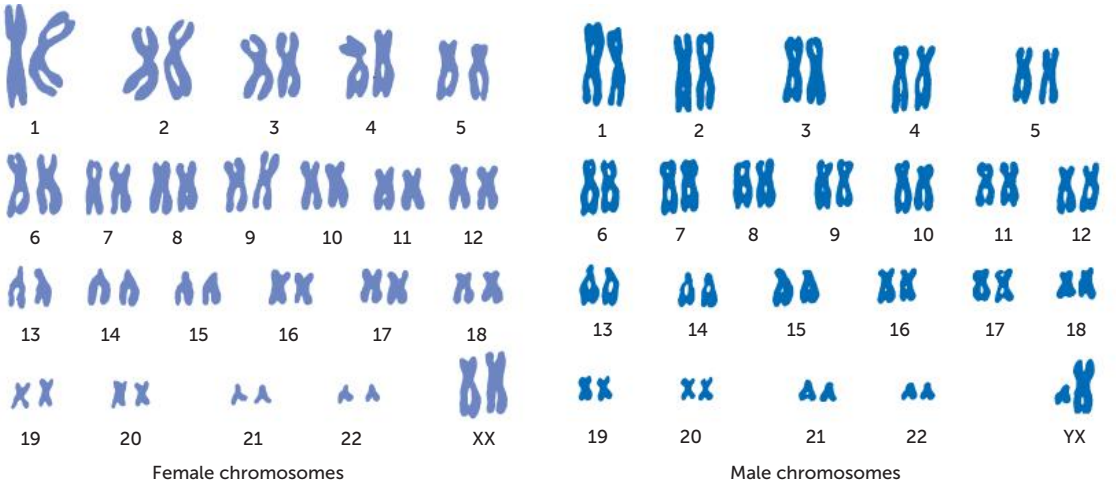


FIGURE 15.21
Human chromosomes: as chromosomes become visible only during cell division, each appears as a double strand ready for division; the strands are joined at one point, so that each double strand is referred to as a single chromosome

Examination of the 23rd pair of chromosomes in males indicated that one of the pair was similar to the chromosomes of the 23rd pair in females, but the other was much smaller. The large chromosome became known as the ***X* chromosome** and the smaller the ***Y* chromosome**. Females, therefore, had two *X* chromosomes, and males one *X* and one *Y*. These are called **sex chromosomes**. The 22 pairs of non-sex chromosomes are called **autosomes**.

Sex determination

It is possible to use logic and a Punnett square to account for the ratio of females to males in the population. All the eggs produced by a female possess an *X* chromosome. On the other hand, half the male's sperm contain an *X* chromosome and half contain a *Y* chromosome. From this information it should be clear that it is the father's sperm that determines the sex of the child. If an *X*-bearing sperm fertilises the egg, the zygote (fertilised egg) will develop into a female; if a *Y*-bearing sperm fertilises the egg, the zygote will develop into a male. As half of the sperm have the *Y* chromosome, there is a 50% chance of the offspring being male. Similarly, as the other half of the sperm have the *X* chromosome, there is a 50% chance that the offspring will be female.

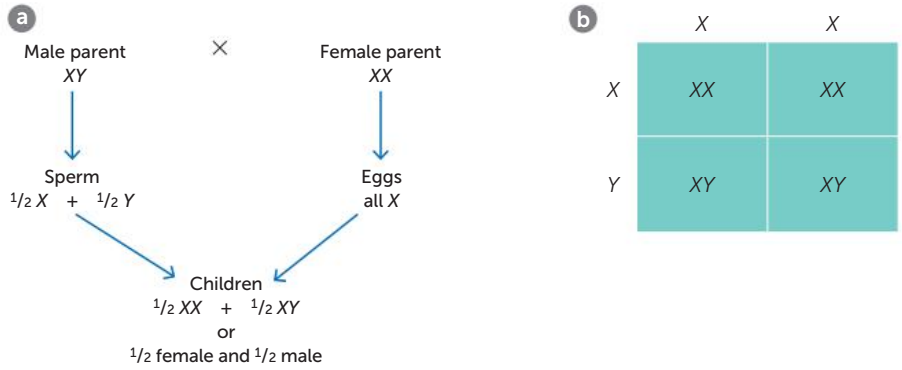
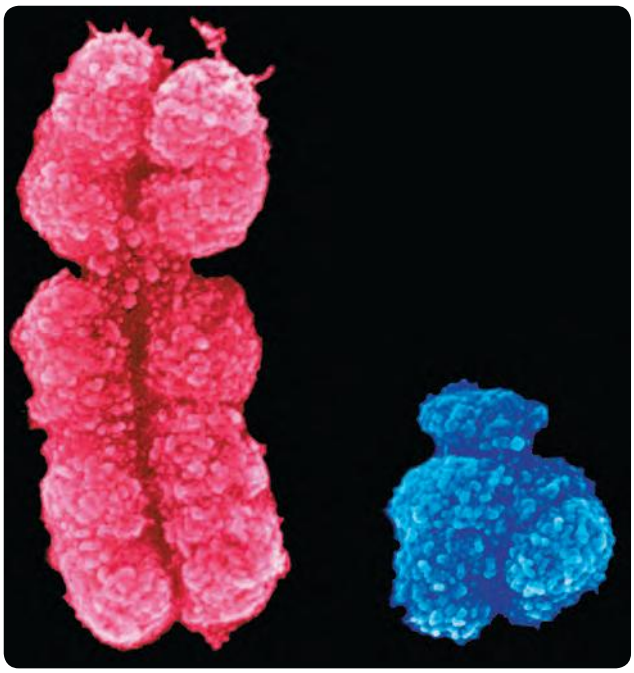


FIGURE 15.22
a Chromosomal basis for sex determination;
b Punnett square for sex determination

Key concept

The sex of a child is determined by the sex chromosomes inherited from the parents. A female child gets an X chromosome from each parent, whereas a male child gets an X chromosome from the mother and a Y chromosome from the father.

Sex-linked characteristics



Alamy Stock Photo/Science Photo Library

FIGURE 15.23 Scanning electron micrograph of an *X* chromosome (red) and a *Y* chromosome (blue)

As you can see in Figure 15.23, the *Y* chromosome is very small compared with the *X* chromosome. Therefore, the *Y* chromosome cannot have the same number of genes in it as the *X* chromosome. This means that most of the genes in the *X* chromosome will lack matching alleles in males. For females, however, the normal pairing of alleles will exist as they have two of the *X* chromosomes.

When characteristics located on the *X* chromosome are studied, it is found that the pattern of inheritance is different in the two sexes. Characteristics that show different patterns in the two sexes are called **sex-linked characteristics** or ***X*-linked characteristics**. When writing the genotypes of sex-linked characteristics, the sex chromosomes are written as capital letters and the allele is written as superscripts for the *X* chromosome. For example, an *X* chromosome with a recessive allele would be represented by X^a.

Sex-linked characteristics may be dominant or recessive. The dominant form is much rarer. They are observed more commonly in females than in males, possibly due to hemizygous males being so severely affected that they do not survive. An example of a dominant sex-linked condition is Rett syndrome. Two common recessive sex-linked traits are red–green colour blindness and haemophilia.

Red–green colour blindness

The ability to discriminate between the colours red and green is controlled by a gene located in the *X* chromosome. Individuals who are unable to distinguish between the two colours possess the recessive allele of this gene. The possible genotypes and phenotypes for **red–green colour blindness** are shown in Table 15.4.

TABLE 15.4 Possible genotypes and phenotypes for red–green colour blindness

GENDER	GENOTYPE	PHENOTYPE
Female	X^BX^B (homozygous dominant)	Normal vision
	X^BX^b (heterozygous)	Normal vision
	X^bX^b (homozygous recessive)	Red–green colour blindness
Male	X^BY (hemizygous dominant)	Normal vision
	X^bY (hemizygous recessive)	Red–green colour blindness

9780170449090

A recessive allele is able to be masked by the presence of a dominant allele in females. However, this is not possible in males; therefore, the frequency of colour blindness is higher in males than in females.

The children of a colour-blind man and a woman homozygous for normal vision would all have normal vision. However, the daughters could produce children who are colour blind, since they are carriers for colour blindness as they carry the recessive allele without showing the phenotype. This is shown in the Punnett square in Figure 15.24a.

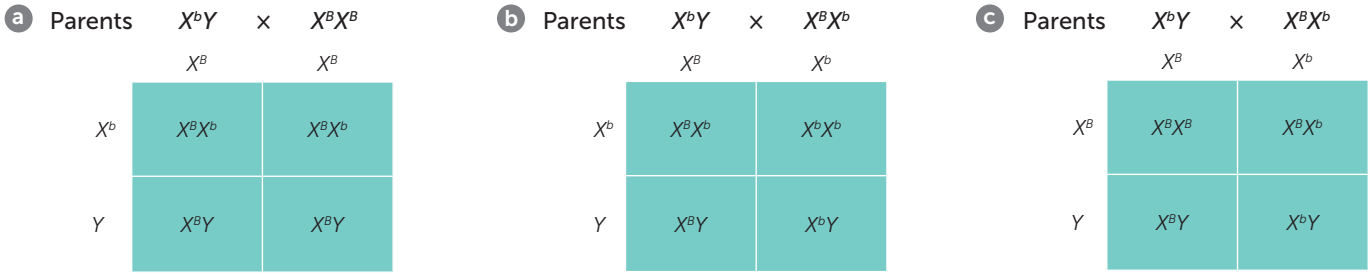


FIGURE 15.24 a Punnett square for a homozygous, normal vision female and a colour-blind male. All the children will have normal vision; however, the female children will be carriers; **b** Punnett square when the parents are a carrier female and a colour-blind male. There is a 50% chance of a female child being colour blind and a 50% chance of a male child being colour blind; **c** Punnett square when the parents are a carrier female and a normal vision male. There is a 0% chance of a female child being colour blind and a 50% chance of a male child being colour blind

As there is no allelic counterpart for males with sex-linked traits, the term 'hemizygous' is used instead of 'homozygous' or 'heterozygous'. Therefore, a colour-blind man is **hemizygous** for the recessive allele.

Haemophilia

Haemophilia is another sex-linked characteristic. It is a relatively rare disease in which the blood clots slowly or not at all. The defective allele is recessive to that controlling normal clotting of the blood and is carried on the *X* chromosome. Males, therefore, can be either normal or haemophiliacs, as they have only one *X* chromosome. Females can be homozygous normal; heterozygous, and therefore carriers of the condition; or haemophiliacs. This last case is extremely rare.

The pattern of inheritance for haemophilia is similar to that already studied for red–green colour blindness. Haemophiliac fathers pass the recessive gene to their daughters. Carrier mothers may pass a defective gene to their sons, who will be haemophiliacs, or to their daughters, who will then also carry the gene. The most famous family pedigree for haemophilia is that of the European royal families descended from Queen Victoria.

Sex-linked inheritance
Use the animation on this website to investigate sex-linked inheritance.

Key concept

Colour blindness and haemophilia are sex-linked characteristic due to a recessive allele on the *X* chromosome.

Patterns with sex-linked inheritance

Knowledge of sex determination and Mendelian inheritance allows us to understand the patterns that occur in sex-linked inheritance, for the following reasons:

- A son's *X* chromosome must come from his mother, as he got his *Y* chromosome from his father. Therefore, a father cannot pass a sex-linked trait on to a son. This also means that if a mother has a recessive sex-linked trait, then any sons will also have the trait.
- A father can only give his *X* chromosome to a daughter. Therefore, if a daughter has a recessive sex-linked trait, then her father must also have it as he cannot be a carrier.
- Females may be carriers of a recessive sex-linked trait. If an unaffected mother has an affected child, then she must be a carrier.
- Two people without a recessive sex-linked trait can have a son with the trait, but not a daughter.

Figure 15.25 shows a pedigree for a recessive sex-linked trait.

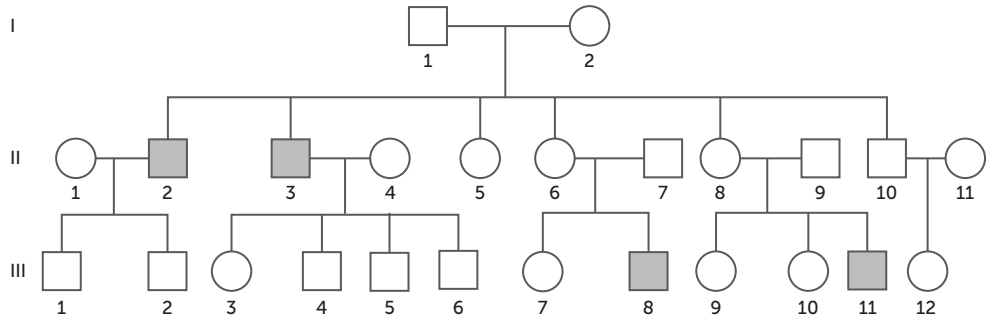


FIGURE 15.25 Typical pedigree for a family showing a recessive characteristic linked to the *X* chromosome

Looking at the pedigree, and knowing that it is for a recessive sex-linked characteristic, we can infer the following:

- Affected individuals (II2, II3, III8 and III11) must be X^hY.
- All other males are unaffected, and therefore must be X^HY.
- Mothers of affected individuals must carry the recessive allele; hence, their genotypes must be X^HX^h.
- Any daughters of affected males must have the recessive allele. As they are unaffected, their genotype must be X^HX^h.
- All other females must have a dominant allele. However, it is not possible to know whether they are homozygous dominant (X^HX^H) or heterozygous (X^HX^h).

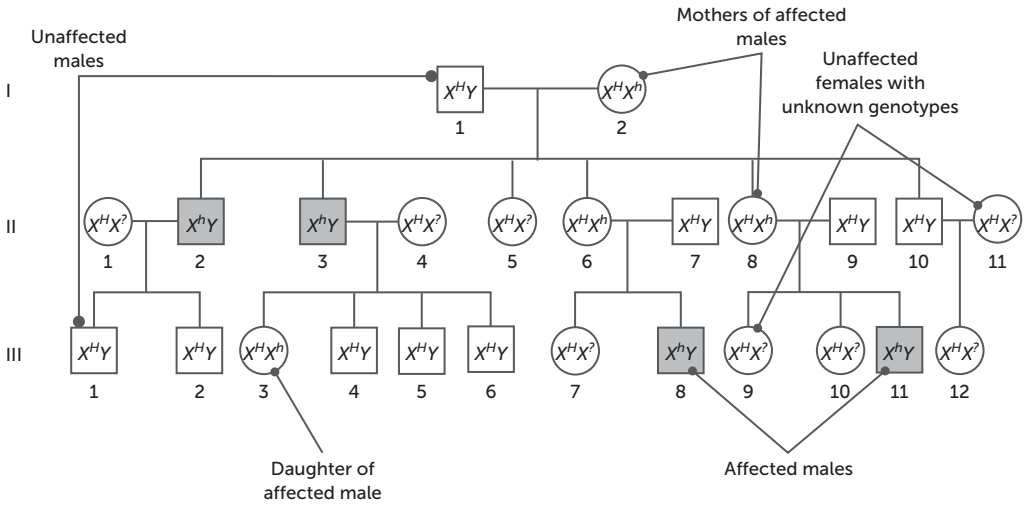


FIGURE 15.26 The pedigree from Figure 15.25 with the genotypes shown

Questions 15.4

RECALL KNOWLEDGE

1 State the number of:
   a autosomal chromosomes in a somatic cell
   b sex chromosomes in a somatic cell
   c autosomal chromosomes in a human gamete
   d sex chromosomes in a human gamete
   e homologous pairs in a somatic cell.
2 Draw a Punnett square to confirm that there is a 50% chance of a child being male.
3 State the sex chromosomes of a:
   a female
   b male.
4 Is the X or Y chromosome longer?
5 Describe the symptoms of red–green colour blindness.
6 Explain why males are referred to as 'hemizygous' when referring to sex-linked characteristics.

APPLY KNOWLEDGE

7 It could be said that the father determines the sex of the child. Discuss whether this statement is true.
8 Explain why sex-linked characteristics have an allele on the X chromosome but not on the Y chromosome.
9 A colour-blind father has a child with a female with normal vision. What is the chance of them having:
   a a child with normal vision?
   b a colour-blind daughter?
   c a son with normal vision?
10 Explain why haemophilia is much more common in males than in females.
11 If a daughter has a sex-linked disorder, her father must also have it. Justify this observation.

15.5 OTHER TYPES OF INHERITANCE

The examples that we have looked at so far were for traits that are determined by a single gene with two alleles – a dominant one and a recessive one. However, not all characteristics have such a simple inheritance. Some other possibilities include variation in the degree of dominance of an allele and the number of alleles of a gene.

Co-dominance

Co-dominance is a situation where two or more alleles are equally dominant. This means that, if both the alleles are present, they will both be observed. One example in humans is the B^M and B^N alleles for the M and N antigens in human blood. Neither allele is dominant to the other, and so three phenotypes are possible:

- type M blood–genotype B^MB^M
- type N blood–genotype B^NB^N
- type MN blood–genotype B^MB^N.

During crosses between various blood types, the identity of the separate alleles is maintained. However, as neither allele is dominant to the other, heterozygous individuals have *both* characteristics because they have both antigens.

The conventions used for representing alleles are outlined in Table 15.5.

TABLE 15.5 Conventions for representing alleles

General conventions	**DOMINANT** alleles are represented by **UPPER-CASE** letters
	recessive alleles are represented by **lower-case** letters
	Do not use letters where upper and lower case are not easily distinguished (e.g. *Ww*, *Ss*, *Cc*).
Co-dominance	**CO-DOMINANT** alleles are represented by **UPPER-CASE** letters
	Use the same letter for each gene; use superscripts of different capital letters for co-dominant alleles (e.g. I^A, I^B).
Sex-linked inheritance	For ***X*-linkage** the *X* and *Y* chromosomes **must** be shown; use superscripts for the alleles (e.g. X^H).

9780170449090

Multiple alleles

Sometimes there are more than two alleles for a particular characteristic. In such cases, they are called **multiple alleles** and the position of that gene on a chromosome is called **multiple allelic**. An excellent example of multiple alleles is seen in the way ABO blood groups are inherited in humans.

A person may belong to blood group A, group B, group AB or group O. This is known as the **ABO blood group system**. Blood groups are inherited, and the ABO blood grouping system is based on the fact that an individual can possess any two of three alternative alleles: I^A, I^B or i. These three alleles are found at the same position on the long arm of chromosome number 9. Blood groups within the ABO system are determined by the inheritance of these alleles, which are responsible for two different antigens found on the membranes of red blood cells. The allele that causes production of the antigen A is usually represented by I^A, and the allele that causes the production of the antigen B is represented by I^B. The third allele does not produce detectable amounts of either antigen A or B and is represented by i.

Alleles I^A and I^B are co-dominant – that is, neither allele is dominant to the other and both characteristics occur in the heterozygote. The allele i is recessive to both I^A and I^B. Its effect is masked by the alleles I^A and I^B.

As allele I^A is dominant to i, the genotypes I^AI^A and I^Ai both produce blood group A; that is, the person has only antigen A. Similarly, I^BI^B and I^Bi both produce blood group B and the person has only antigen B.

The alleles for I^A and I^B are co-dominant, and so the genotype I^AI^B forms a third phenotype, AB, a person with *both* antigens. The homozygous recessive, ii, forms the fourth phenotype, group O blood, where neither antigen is produced. Table 15.6 summarises the phenotypes and genotypes of the ABO blood group system.

Knowledge of the inheritance of blood groups was sometimes used to determine the parentage of a particular child. For example, it would be impossible for a child whose blood group was AB to have parents whose blood groups were A and O, as neither parent has an I^B allele to pass on to the child.

TABLE 15.6 Genotypes and phenotypes of the ABO blood group system

GENOTYPE	PHENOTYPE	BLOOD GROUP
I^AI^A I^Ai	Antigen A	A
I^AI^B	Antigen A and antigen B	AB
I^BI^B I^Bi	Antigen B	B
ii	Neither antigen A nor antigen B	O

Key concept

The ABO blood group system is controlled by three alleles, two of which are dominant while the other is recessive. Therefore, this is an example of a multiple allelic, co-dominant trait.

Questions 15.5

RECALL KNOWLEDGE

1. Describe co-dominant inheritance.
2. The alleles for the ABO blood group are represented by I^A, I^B and i. Explain why:
   a. the letter i is used for all alleles
   b. two alleles use a capital I but the other is lower case
   c. the letters A and B are used.
3. State the genotype for someone with type MN blood.
4. What is the blood type of someone with an I^Ai genotype?

APPLY KNOWLEDGE

5. Justify the ABO blood group being an example of both co-dominance and multiple allele inheritance.
6. A child's blood type is AB. List all the possible genotypes of the parents, including Punnett squares to support your answers.
7. Explain why a parent with type O blood can have a child with type A blood, but not type AB.

9780170449090

15.6 GENETIC COUNSELLING

Today, most women in Australian society are aware of the risks of producing a baby with a birth defect. During pregnancy, most women take special care of their health and avoid alcohol, cigarettes and other drugs, to ensure that their baby is as healthy as possible. However, if a couple have already produced a child with a birth defect, they will naturally be very anxious about the possibility of the same thing happening in later pregnancies. In other cases, one partner may have a genetic defect and the couple will be concerned that the problem could occur in their children. Other couples may have close relatives who have inherited diseases or have given birth to children with such disorders. In all these cases, the people concerned may seek additional information about the risk of the inherited disorder occurring in their children.

Genetic counselling

One form of advice is **genetic counselling**. By examining the incidence of a disorder in the family tree, the probability that a particular condition will occur can sometimes be determined. The couple can then decide whether to risk having a baby with the inherited disorder.

For example, if a couple with no history of genetic disorders in either family had a child with an autosomal recessive condition such as thalassaemia, what would be the probability that their next child would inherit the same condition? A genetic counsellor would tell them that there was a one-in-four chance of each of their subsequent children having thalassaemia. Should they decide to have another child under these circumstances? As you saw in Chapter 14, there are now procedures available that can detect the presence of many genetic disorders in the foetus before birth. If a genetic disorder were diagnosed, the couple would then have the option of terminating the pregnancy. Although genetic counselling and modern diagnostic techniques can alleviate much suffering, the responsibility for decision making must still lie with the individuals concerned. The decision to risk having child with a genetic disorder, or to terminate a pregnancy, is not made lightly.

Profiling techniques

In the past, genetic counselling relied largely on information from family pedigrees to determine the likelihood of a couple having a child with a certain genetic disorder. Advances in biotechnology have resulted in the development of techniques to identify the unique genetic make-up of individuals. For some inherited conditions, this has enabled couples to know with more certainty the chances of having an affected baby. As with many techniques in medical science, although there are tremendous benefits, there are also risks and ethical concerns.

A person's DNA is so distinctive that it can be used as a means of identification. In the late 1960s, scientists developed techniques using special enzymes to cut the DNA at specific base sequences, leaving pieces of various lengths. The length of these pieces varies distinctively from one person to another. However, it was not until 1984 that a breakthrough enabled the technique to be refined. The DNA pieces were placed on a bed of semi-solid gel and an electric current was passed through the gel via electrodes located at each end, a technique called **electrophoresis**. The DNA, which is negatively charged, moves through the gel towards the positive electrode. The smaller DNA pieces move faster than the larger ones, resulting in a pattern of bands that looks similar to the barcodes on products sold in supermarkets. This banding pattern is an individual's **DNA fingerprint**, often called a **DNA profile**.

DNA fingerprints are frequently used in tracing ancestry and in forensic science. They are also useful in the identification of carriers for hereditary diseases. Using gel electrophoresis, a person who carries an allele that may cause a hereditary disease, such as cystic fibrosis or Huntington's disease, can be identified, as can a newborn baby who will develop the disease. In some cases, the fact that a person has a particular allele does not automatically mean that they have the disease, or will even develop it. A recently discovered allele has been shown to increase an individual's risk of colon cancer.

The history of DNA profiling
This BBC website has a brief article about the development of DNA profiling.

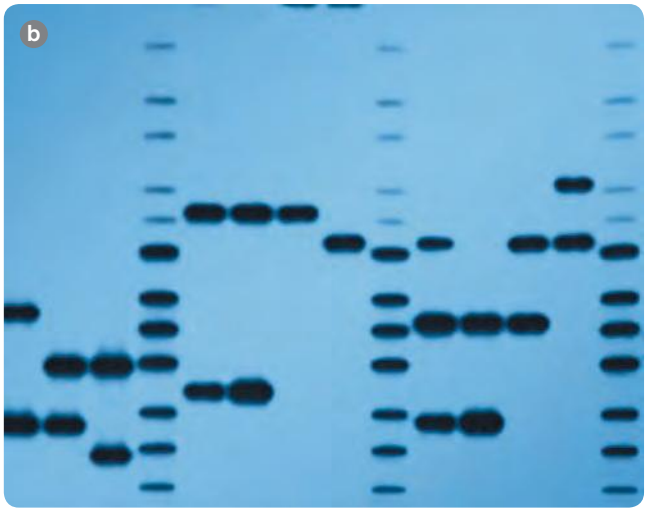


FIGURE 15.27 a Adding a sample to the gel electrophoresis; **b** A DNA fingerprint

DNA profiling enables this allele to be identified, and so a person with the allele can then have regular medical examinations, even though they may never develop colon cancer.

DNA profiling enables many inherited diseases to be detected at an early age. Early diagnosis provides a greater chance that the condition can be effectively treated and possibly cured. People with a history of an inherited disease in the family can use a DNA profile to determine their risk of having an affected child.

DNA profiling is also used in determining parentage, and an example of how it can be used is shown in Figure 15.28. The DNA fingerprints of a family are shown. Emma and James are biological brother and sister. They do not have the same DNA profile but there are similarities. You can also see that each of them has some bands in their profiles that are the same as bands in each of their parents' profiles. Yet, neither has DNA that is identical to one parent or the other. Liam is adopted. His DNA has no bands that are the same as his adoptive parents'.

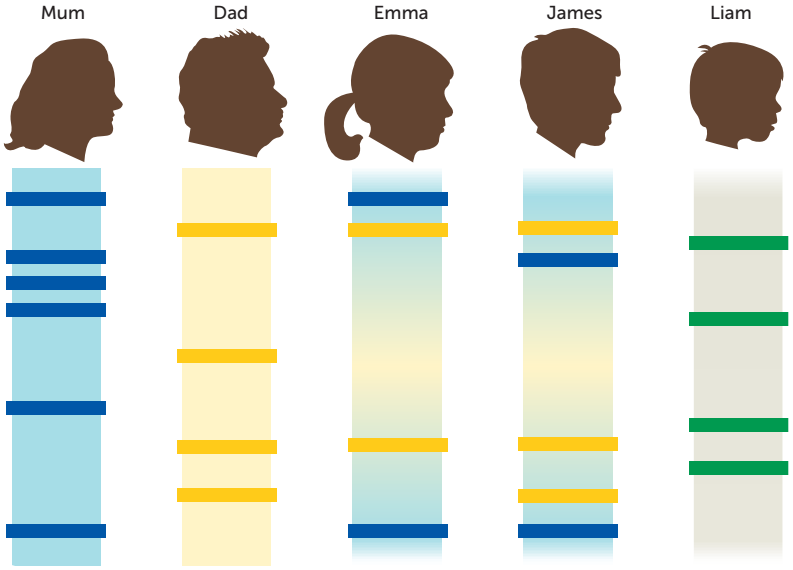


FIGURE 15.28 DNA fingerprints of a family

In comparing DNA fingerprints, scientists use markers. A **marker** is a segment of DNA with known characteristics. These segments of DNA do not code for a protein, but they contain short sequences where the same pattern is repeated a number of times. Because the number of repeats in these sequences is inherited, they make useful points of comparison in genetic testing.

To determine a link between two individuals, specific markers on the DNA strand are studied. Scientists look for the number of repeats at each marker, examining between 20 and 40 markers. If 10 different markers on different chromosomes are examined, there is only a one in a million chance that two people will have the same number of

FIGURE 15.29 A DNA fingerprint used to help establish the identity of a child's father. The fact that the DNA bands match does not necessarily mean that the person is the father; however, a mismatch would definitely exclude that person from being the father

repeats, except for identical twins. The more markers that match, the more likely it is that the two individuals are related.

Each of your parents received their DNA from each of their parents, and so on. DNA can be used to trace ancestry back through your family tree. As DNA is passed down from one generation to the next, some parts remain almost unchanged, while other parts change greatly. These unchanged parts of the DNA molecules provide a link between generations and can be very useful in reconstructing family histories. However, they cannot provide a person with their entire family tree. DNA testing can only tell if two people share a common ancestor. People who marry into a family will have completely different DNA from family members who are directly related.

Ethical and social issues

Genetic profiling and its applications raise many ethical and social issues. Ethics is the set of moral principles or values that are held by the majority of Australian society. Profiling has the potential to breach this unwritten code of values. The way in which society uses profiling also presents problems.

Genetic profiling provides a complete set of genetic information about a person – their **genome**. It has the potential to provide great benefits for individuals and for providers of health care. A person's genome could be analysed during infancy, or even before birth, and stored electronically to be used later if required, but this raises significant questions. Who does the statement of the genome belong to – the individual, the laboratory that carried out the tests, the medical authorities who may wish to use the record, or some other person or group? Who will be allowed access to the stored record? How will the person's right to privacy be assured? What happens if the data is stolen or misused?

On the other hand, there are many potential benefits from having access to one's genome. Future diseases may be predicted so that effective intervention and early treatment may be possible. This is particularly the case with single-gene disorders that can be detected during embryonic development. If a genome indicates the presence of a single-gene disorder, this knowledge could be passed on to relatives who may be at risk of developing the disorder or may have children with the disorder.

Knowing one's genome could enable a more proactive approach by encouraging more personal involvement in decision making about health matters. However, in many cases when potential for disease is discovered, there is no way that development of the disease can be prevented and no treatment when the disease does develop. If testing does show that a disease will develop, it cannot indicate the age of onset of the disease or its severity. The question then arises as to whether testing is in a person's best interest. It may lead to unnecessary anxiety, and possibly stigma, and it may change the person's perception of wellness.

In many cases, genetic testing will yield little definitive information. Diseases like diabetes, heart disease and kidney disease and many cancers are caused by multiple interacting factors. A person's genome is just one of these factors, and genetic profiling cannot possibly assess the risk of developing such a disease. Another reason that genetic testing results may be unreliable is that a person's environment can modify the expression of a gene, a situation known as epigenetics (see Chapter 9).

The use of DNA profiling in determining parentage and ancestry also raises many ethical considerations. The supposed parents must be willing to provide a DNA sample, but there have been cases of samples being obtained without a person's knowledge. An individual should be able to control who receives information about their genetic make-up.

DNA testing to determine paternity has become widely sought after in many countries. For example, studies in Italy indicate that the number of children not being raised by their biological father may be as high as 15%. Cases of divorce have arisen as fathers may choose not to continue bringing up a child they had thought was theirs prior to doing a DNA test.

Personal genetic profiling services are now available in Australia. If you search the Internet you will find many laboratories that, for a fee, will analyse a saliva sample and provide you with an estimated risk of developing certain diseases.

9780170449090

Profiling has the potential to cause discrimination. In Australia, we all pay about the same for health insurance regardless of genetic profile and family or personal health history. If misused, genetic profiles could result in people paying for health cover according to their risk of disease. Life insurers in Australia cannot insist on genetic testing before they provide insurance cover, but they can demand to see a genetic profile if one has already been carried out. Insurance premiums could then be adjusted accordingly. The same kind of discrimination could occur with employment – employers could give preference to potential employees who have some desired genetic profile.

Some of the advantages and disadvantages of genetic profiling have been discussed. As with all issues involving ethics, it is up to each person to consider the arguments for and against the issue and then decide where they stand.

Key concept

Genetic counselling and genetic profiling can be used to identify the risk of genetic disorders. However, there are ethical factors to consider when deciding whether to undertake these options.

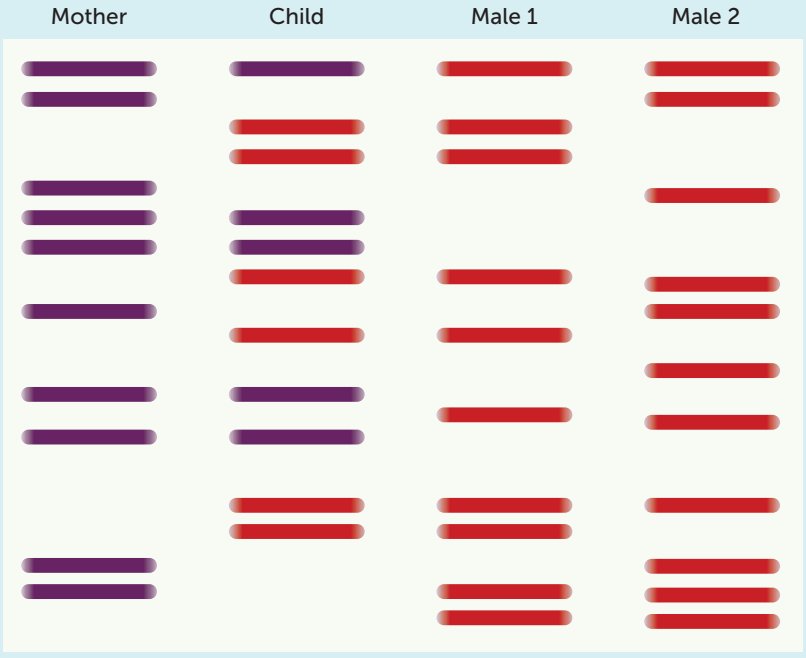
Questions 15.6

RECALL KNOWLEDGE

1 Define 'genetic counselling'.
2 Name the technique used to determine a DNA fingerprint.
3 List some situations where genetic profiling could be considered to be:
   a a positive option
   b a negative option.

APPLY KNOWLEDGE

4 Explain why some couples may choose to seek genetic counselling prior to starting a family.
5 DNA profiling can be used to identify paternity. Use the DNA profiles below to identify which male is the father of the child.



6 Explain why 20–40 markers are studied in DNA profiling, rather than just one.

CHAPTER 15 ACTIVITIES

ACTIVITY 15.1 Investigating Mendelian genetic principles in Martians

Martians are an imaginary group of people from the red planet, Mars. Their skin colour is determined by two alleles: one for red skin colour and one for white skin colour. Red is dominant to white on Mars. In this activity, we will investigate whether Martians follow the principles of Mendelian genetics by simulating a cross between two heterozygous Martians.

You will need

For each pair:

- 2 containers (2-litre ice-cream containers work well)
- 20 red beads or counters to simulate the dominant red allele (R) in each gamete
- 20 white beads or counters to simulate the recessive white allele (r) in each gamete
- felt pen, tally sheet, pencil

What to do

1. Label one container 'Male parent' and the other 'Female parent'.
2. In each container, place 10 of the red beads (gamete with *R* allele) and 10 of the white beads (gamete with *r* allele).
3. Prepare a tally sheet similar to the one below.

	GENOTYPES IN THE MARTIAN OFFSPRING			
	RR	Rr	rR	rr
1				
2				
3				
... 10				

4. Shake the containers well. Draw out one bead (gamete) from each container in turn and place a tick in the relevant box on the tally sheet to show the combination of alleles in the offspring.
5. When you have completed 10 draws, place the beads back into the container. Your partner should repeat steps 1 to 4. Together you should now have two completed tally sheets.

Studying your data

1. Because the first three columns all contain the dominant allele (*R*), individuals with these genotypes will all appear red. Tally up the number of red offspring.
2. Individuals with the genotype *rr* will appear white. How many white offspring do you have?
3. What is the ratio of the phenotypes, red to white?
4. Combine your data with that of the other groups in the class to obtain a larger sample. What is the ratio now?

Interpreting your data

5. Has this activity shown that inheritance of skin colour in Martians follows the principles of Mendelian genetics?
6. How close were your results to the expected result of 3:1?
7. Was the ratio calculated by combining all groups in the class closer to the expected? Explain why this was the case.

9780170449090

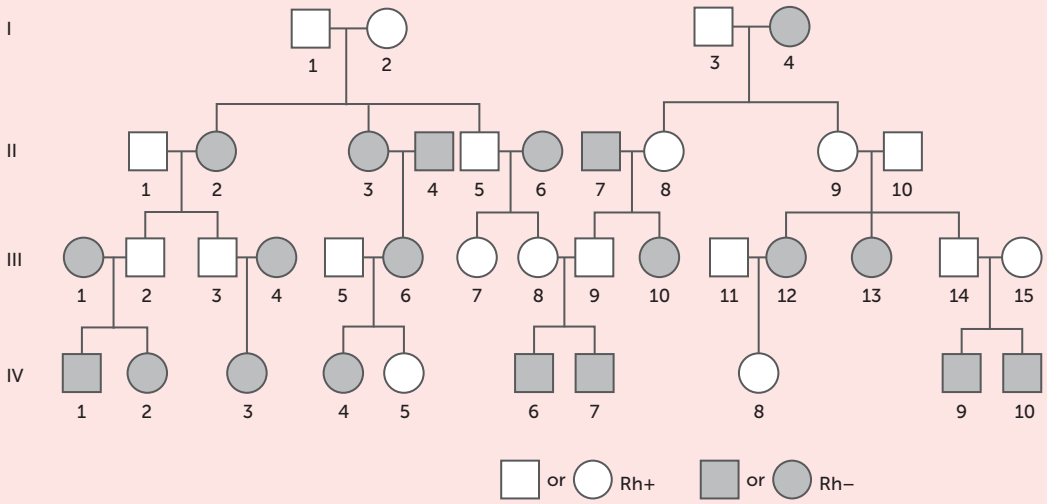
ACTIVITY 15.2 Examining pedigrees

Pedigree 1

The inheritance of Rh blood groups in humans follows the laws of simple dominance. The letters 'Rh' were used because it was from experiments in 1939 with the blood of rhesus monkeys that the blood groups were first identified. There are two types: Rhesus-positive (Rh+) and Rhesus-negative (Rh–). In Australia, about 85% of the population is Rh+ and 15% Rh–.

What to do

Study the following pedigree of a family in which some members are Rh–. The Rh+ trait is determined by a dominant allele, and people who are Rh+ are shown by open symbols. Shaded symbols represent Rh– people. Determine the genotypes of all the individuals shown.



Interpreting your results

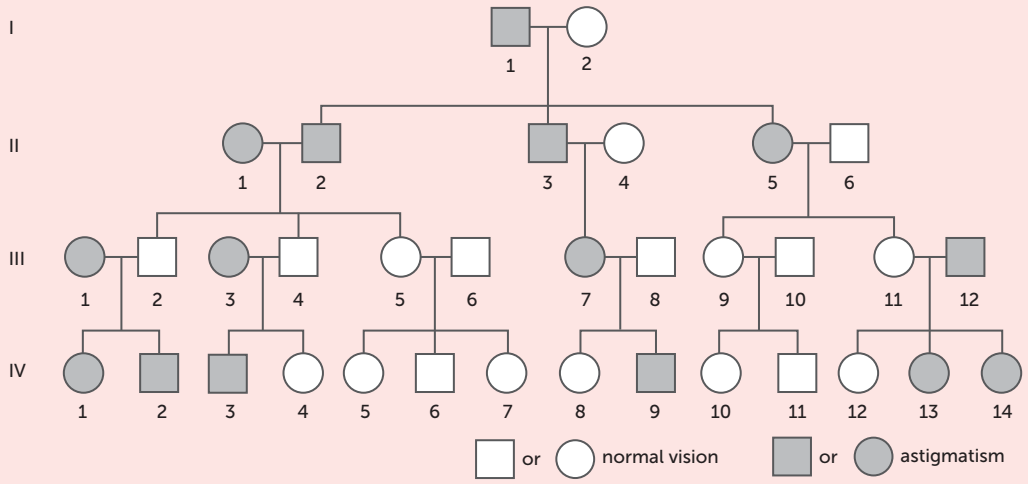
1. Can you be absolutely certain about the genotypes of all individuals in the first generation? Give reasons for your answer.
2. What are the genotypes of the male (III 5) and the female (III 6) in the third generation who married and produced two daughters? Describe the genotypes using both words and the appropriate letters. Explain why you can be certain of their genotypes.
3. Do the two Rh+ females in the fourth generation (IV 5 and IV 8) have the same genotype? Explain your answer.
4. Is the Rh blood group controlled by a gene on an autosomal or an *X* chromosome?

Pedigree 2

In many families, the visual defect astigmatism is an inherited condition. The pedigree on the following page shows one such family.

What to do

Carefully examine the pedigree and work out the genotypes of all individuals shown. Is there any member of the family about whose genotype you are uncertain?



Interpreting your results

Is astigmatism in the family pedigree above dominant or recessive? Give reasons for your answer.

ACTIVITY 15.3 Studying a family with Huntington's disease

Huntington's disease is described as an inherited disorder that results in lack of control over muscles and progressive mental deterioration to the point where sufferers are unable to look after themselves. The symptoms rarely appear before 40 years of age and by that time individuals with the disorder may have passed the allele for the condition on to their children. Huntington's disease is transmitted by a dominant allele.

The following paragraphs describe a family in which Huntington's disease has occurred.

Jennifer is 45 years old and has just developed the symptoms of Huntington's disease. Her father, James, is 70 years old and is hospitalised with the disorder, but her mother, Anne, two years younger than her father, does not have the condition. Jennifer's husband, John, also 45 years old, does not have Huntington's disease, and there is no history of the condition in his family. Jennifer's older brother, Malcolm, does not have the disease.

Jennifer and John have two children, Andrew (25 years old) and Michele (21 years old). Michele is married to Tony, who is the same age as her brother, and she has just given birth to their first child, called Darren. There is no history of Huntington's disease in Tony's family.

What to do

Construct a pedigree to show all the individuals in the family. Indicate the individuals who have Huntington's disease by shading the relevant circles or squares.

Interpreting the family tree

1. Write down the possible genotypes of James, Anne, Jennifer and John. Explain the symbols you are using.
2. What is the probability that Michele has inherited Huntington's disease? Using a Punnett square, set out the cross between Michele's parents in full.
3. Is there any possibility that Darren has inherited the disease? Explain, using a Punnett square to set out the cross between his parents in full.
4. Is the gene that determines whether a person has Huntington's disease located on an autosomal or an X chromosome? Explain your answer.

9780170449090

ACTIVITY 15.4 Investigating patterns of inheritance in heterozygous barley seeds

Barley (*Hordeum vulgare* L.) was one of the first cultivated grains. A member of the grass family, barley is now grown in more than 100 countries. Barley has 14 chromosomes and self-pollinates asexually to reproduce. A single gene with two alternative alleles controls pigmentation in barley. The dominant allele results in a gene-dominant phenotype and green pigmentation. The other allele produces no pigmentation, resulting in a white (or albino) recessive phenotype. In the heterozygote, the dominant expression of green pigment masks any expression of the allele coding for no pigment (albino).

Aim

To perform a monohybrid cross and predict phenotypic ratios.
Time requirement: 20 minutes

You will need

25 seeds of genetically selected barley; filter paper; disposable plastic Petri dish; plastic pipette; forceps

Risks

WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
Some people may be allergic to particular seeds	Do not eat the seeds. Wash hands thoroughly after handling seeds.

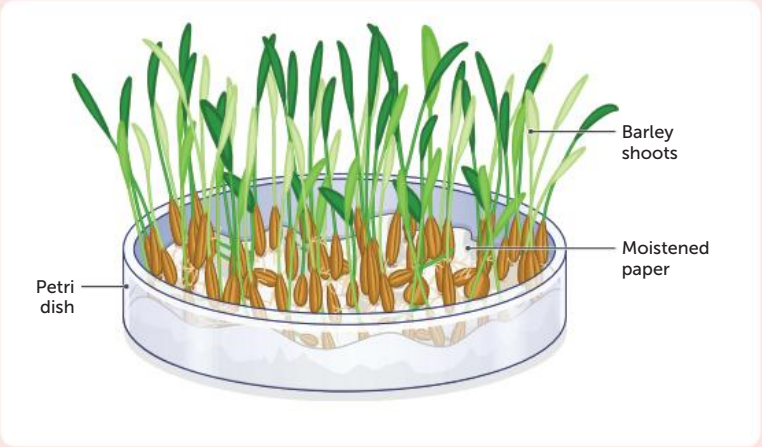
What to do

1. Place a piece of filter paper into the bottom half of each Petri dish. Trim the paper as necessary so that the paper lies flat in the bottom of each dish.
2. Soak the filter paper with tap water using a pipette. Remove or drain any excess water that is not absorbed by the paper.
3. Sprinkle the seeds evenly over the moistened paper in the Petri dish. Ensure the seeds are evenly spread out (approximately 1 cm apart).
4. Place the Petri dish with seeds on a bench with sufficient access to sunlight to keep them at room temperature.
5. Rehydrate the seeds twice a day using a pipette to prevent them from drying out. This process of twice-daily rehydration should continue until the barley seedlings reach a height of 2 cm, which will take approximately one week.
6. Propose a hypothesis of the phenotypic ratio you expect to see using a Punnett square (monohybrid cross), where 'A' represents pigment produced (green, dominant) and 'a' represents no pigment produced (white, recessive albino).

	A	*a*
A		
a		

Predicted ratio:

7. Observe the Petri dishes at the end of one week. Some seedlings will be pale in colour (albino) with little or no green pigment. Other seedlings will have green areas forming. When nearly all the seedlings have germinated, count each seedling as either green or albino. Record your results and contribute your individual results to the class data.



Studying your results

1 Record your individual and class data by copying and completing the table below.

	TOTAL NUMBER OF SEEDLINGS	NUMBER OF SEEDLINGS OF EACH COLOUR	
		Green	Albino
Individual data			
Class data			

2 Calculate the ratio of green seedlings to albino seedlings for:
   a your individual data
   b the class data.
3 How do your individual data compare to the ratio for the class data?

Discussion

1 What is one limitation of this investigation?
2 How would you improve the reliability and validity of the data in this procedure?
3 Was your predicted ratio based on your Punnett square proven correct?
4 Were there any inconsistencies in the results? If so, explain why they may have occurred.
5 Based on your individual and class results, what is the mode of inheritance for green pigmentation in barley?
6 From a visual standpoint, homozygous green barley plants are indistinguishable from heterozygotes. To identify the genotype of an individual plant showing the dominant characteristic, a geneticist undertakes a test cross. Describe a test cross.
7 If presented with 120 seedlings, approximately how many would you expect to be green? Show your working out.

Conclusion

Summarise your findings in this activity, commenting on your hypothesis and the mode of inheritance for pigmentation in barley.

Taking it further

1 In this investigation you have conducted a monohybrid cross. What type of investigations could you conduct to demonstrate a dihybrid cross and a sex-linked cross?
2 Calculate the chi-square value for this experiment. Do your observed frequencies deviate significantly from the expected frequency of this cross?

9780170449090

CHAPTER 15 SUMMARY

- Mendel's principles of inheritance state that inherited characteristics are controlled by genes, and that each gamete receives one set of genes.
- Mendel conducted experiments on garden peas, looking at characteristics such as seed shape, flower position and stem length. He started with plants that were pure-bred for the trait. When two plants pure-bred for the different forms of the trait were crossed, only the dominant trait was shown.
- The principle of segregation states that during the formation of gametes, only one gene for each trait is included.
- Alleles are the different forms of a gene. The dominant allele is represented by a capital letter. The recessive allele is represented by the lower-case letter.
- The genotype is the combination of alleles for a trait. Homozygous genotypes have two of the same type of allele, whereas the heterozygous genotype has both a dominant and a recessive allele.
- The phenotype is the expression of the genotype.
- The offspring of the parents are called the first filial generation (F_1), and their offspring are the second filial generation (F_2).
- Punnett squares are used to model a cross and the possible genotype of the offspring. They can be used to predict the probability of different phenotypes.
- Pedigrees model the phenotypes of family members. Each member is represented by a square (male) or a circle (female), drawn in rows by generations. Connections between generations are shown by vertical lines. Individuals with the trait are shaded.
- Patterns of phenotypes are used to determine the genotypes of individuals and, hence, the pattern of inheritance.
- Single-gene disorders are caused by the inheritance of a single gene.
- For autosomal dominant traits, parents without the trait are unable to have children with the trait.
- Achondroplasia, facioscapulohumeral muscular dystrophy, Huntington's disease and neurofibromatosis are examples of autosomal dominant diseases.
- Huntington's disease is an autosomal dominant disease that causes involuntary movement of the limbs and dementia.
- For autosomal recessive traits, both parents must have a recessive allele to pass on, and the individual must pass on a recessive allele to all children. Individuals with a heterozygous genotype will not show the trait, but are able to pass it on. Therefore, they are called carriers.
- Phenylketonuria and cystic fibrosis are examples of autosomal recessive disorders.
- The 46 chromosomes in humans can be classified as autosomal chromosomes or sex chromosomes: 44 chromosomes (22 pairs) are autosomal and 2 (1 pair) are sex chromosomes.
- Sex chromosomes are either an *X* chromosome or a *Y* chromosome. The *Y* chromosome is much shorter than the *X* chromosome. Females have two *X* chromosomes (*XX*) and males have one of each (*XY*).
- During meiosis, only one sex chromosome goes into each gamete. Every egg will have an *X* chromosome. Half the sperm will have an *X* chromosome and the other half will have a *Y* chromosome.
- If the zygote forms from an egg and a sperm with *X* chromosomes, it will be a female. If the zygote forms from an egg with an *X* chromosome and a sperm with a *Y* chromosome, it will be a male.

9780170449090

- Due to the shorter length of the *Y* chromosome, most of the genes on the *X* chromosome do not exist on the *Y* chromosome. Therefore, males only have one copy of these genes, but females have two. Traits found on the *X* chromosome are called sex-linked characteristics.
- The genotypes for sex-linked characteristics of females may be homozygous dominant (X^BX^B), homozygous recessive (X^bX^b) or heterozygous (X^BX^b).
- The genotypes for sex-linked characteristics of males may be hemizygous dominant (X^BY) or hemizygous recessive (X^bY). Males cannot be heterozygous, and therefore cannot be carriers.
- Red–green colour blindness and haemophilia are sex-linked characteristics.
- Co-dominant alleles are equally dominant. Therefore, if both alleles are present, both are observed. Both alleles are represented by the same upper-case letter, with a superscript representing the alleles.
- The MN blood groups are an example of a trait with co-dominant alleles.
- Characteristics that are controlled by more than two alleles are called multiple allelic.
- The ABO blood group system is an example of a multiple allelic characteristic. Two of the alleles are co-dominant (I^A and I^B) and the third allele is recessive (i). The dominant alleles will lead to the production of antigens. The blood groups are named from the antigens produced. Blood group O is due to homozygous recessive genotype, where neither antigen A nor antigen B is produced.
- Genetic counselling provides advice regarding the probability of a particular problem based on the incidence in a family.
- Genetic profiling can use electrophoresis to separate the DNA to produce a DNA profile. This can be used for forensic science, ancestry research and tracing hereditary diseases. It can allow inherited diseases to be detected before symptoms show, and the probability of parents having a child with the disease to be determined.
- There are ethical considerations for genetic profiling, including providing consent for the DNA to be collected and used, who owns the information, the potential uses of the information and whether the knowledge is beneficial.

9780170449090

CHAPTER 15 GLOSSARY

ABO blood group system A system of classifying blood types according to the antigens on the surface of the red blood cells

Albino An individual who lacks pigmentation, resulting in white skin, white hair and pink eyes (due to the reflection from blood vessels in the eyes)

Allele The alternative forms of a gene that occur at a given point in a chromosome

Autosome A non-sex chromosome

Carrier An individual who carries a recessive allele that is not expressed in their appearance

Co-dominance When contrasting alleles both affect the appearance of an individual and neither is dominant over the other

Consanguineous union The union of two close relatives, usually cousins

Cystic fibrosis A disorder controlled by a recessive allele; it results in chest infections, a lack of digestive enzymes and increased salt loss

Dementia A brain disorder resulting in memory loss, personality changes and impaired reasoning

DNA fingerprint *see* DNA profile

DNA profile The pattern of bands revealed by the process of electrophoresis; also called DNA fingerprint

Dominant trait One of a pair of contrasting characteristics, which is controlled by an allele that is not masked by other alleles

Electrophoresis A technique used to reveal DNA profiles in a banded pattern

First filial generation (F_1 generation) The offspring of the first set of parents

Gene The factor that determines a hereditary characteristic; part of a chromosome

Genetic counselling Advice regarding the risks of genetic disorders in future children

Genome The complete set of genetic information of an organism

Genotype The genetic constitution of an individual

Haemophilia An inherited disorder in which the blood clots slowly or not at all

Hemizygous Having no allelic counterpart; occurs with alleles in the X chromosome in males

Heterozygous Possessing different alleles for a given characteristic

Homozygous Possessing the same alleles for a given characteristic

Huntington's disease A hereditary disease, the symptoms of which seldom appear before 40 years of age, characterised by occasional involuntary flailing movements of the arms and legs

Hybrid An offspring that is the result of a mating between individuals of two different genetic constitutions

Marker A segment of DNA with known characteristics; can be used to compare DNA fingerprints

Monohybrid cross A mating between individuals in which only one pair of contrasting characteristics is being considered

Multiple alleles Three or more alternative forms of a gene for a characteristic; although there may be many alternative forms, each person has only two of the alternatives

Multiple allelic Traits governed by three or more alleles

Pedigree A family tree

Phenotype The appearance of an individual as determined by their genetic constitution

Phenylketonuria (PKU) An inherited disease resulting in damage to the growing brain, and thus extreme intellectual disability; also a tendency towards epileptic seizures and a failure to produce normal skin pigmentation

Principle of segregation The principle that the alleles for a trait are separated during the formation of gametes

9780170449090

Progeny Offspring

Punnett square A grid used to predict the probability of genotypes of offspring

Pure-breeding The production of the same characteristic in each succeeding generation when individuals are bred among themselves; homozygous

Recessive trait One of a pair of contrasting characteristics, which is controlled by an allele that is masked by dominant alleles

Red–green colour blindness A sex-linked recessive trait that causes the colours red and green to be perceived as identical

Second filial generation (F_2 generation) The offspring from crossing the first filial generation

Sex chromosomes The pair of chromosomes that determines the sex of an individual

Sex-linked characteristic A characteristic determined by genes carried on the X chromosome

Single-gene disorder A disorder caused by the inheritance of a single defective gene

Trait The characteristics due to the genetic constitution of an individual

X chromosome One of the two sex chromosomes; it contributes to the determination of the sex of an individual

X-linked characteristic *see* sex-linked characteristic

Y chromosome One of the two sex chromosomes; it contributes to the determination of the sex of an individual

9780170449090

CHAPTER 15 REVIEW QUESTIONS

Recall

1 Define the following terms:
   - a pure-breeding
   - b progeny
   - c hybrid
   - d dominant
   - e recessive
   - f co-dominant
   - g carrier
   - h hemizygous
   - i first filial generation.

2 Briefly describe what is meant by the principle of segregation.

3 Describe the difference in appearance between the X and Y chromosomes.

4 a What are autosomes?
   - b How many autosomes occur in:
     - i each normal human cell,?
     - ii each sperm or egg?

5 a What are sex-linked characteristics?
   - b Give examples of such characteristics.
   - c Why are sex-linked characteristics also called X-linked characteristics?

6 List five rules that must be observed when constructing a pedigree.

7 Describe the pattern of inheritance of the following disorders:
   - a cystic fibrosis
   - b red–green colour blindness
   - c Huntington's disease
   - d phenylketonuria.

8 Phenylketonuria is one genetic disorder discussed in this chapter. Briefly outline:
   - a the symptoms caused
   - b how it can be identified in newborn infants
   - c the treatment that is given.

9 a Describe what is meant by the term 'DNA profile'.
   - b List benefits that have arisen for those with hereditary disease, from the use of DNA profiling.
   - c How has DNA profiling contributed to determination of parentage and ancestry?
   - d State one ethical consideration with genetic profiling.

Explain

10 Using examples, distinguish between:
   - a homozygous and heterozygous
   - b phenotype and genotype
   - c allele and gene.

11 Use an example to explain co-dominance.

12 Explain how the sex of a child is determined at the time of fertilisation.

13 Explain how multiple alleles are important in blood groups.

14 Explain why a father with an X-linked condition is not able to pass the characteristic to his sons.

15 Describe what is meant by 'genetic counselling', and discuss how it may assist people in deciding whether to have a child or to continue with a pregnancy.

Apply

16 Using your own family as an example, explain the difference between a first filial generation and a second filial generation.

17 a What is probability?
   - b If two people with normal skin colouring, each with the recessive allele for albinism, have a child, what is the probability of the child being an albino?

18 Why do couples who are first cousins have a slightly higher risk of having a child with an inherited disorder than unrelated couples?

9780170449090

19 In garden peas, round seed shape is dominant to wrinkled seed shape. Pure-breeding round seed plants were crossed with pure-breeding wrinkled seed plants. Determine the expected genotypes and phenotypes of the F_1 and F_2, and the expected proportions.

20 In humans, normal melanin production is dominant to albino, which produces white hair and pink eyes. The first child born to a married couple with normal pigmentation is an albino. Calculate the probability that the second child will also be an albino. Give a clear explanation for your results.

21 In guinea pigs, black fur colour is dominant over white fur colour. How could an animal breeder test whether a black guinea pig is homozygous or heterozygous?

22 In humans, free earlobes are dominant over attached earlobes. A woman heterozygous for free earlobes marries a man with attached earlobes. Use a Punnett square to determine their chance of producing children with attached earlobes.

23 In many families, a Roman-shaped nose is dominant to a straight nose. If a man from a family pure-breeding for a Roman nose has children with a woman from a family pure-breeding for a straight nose, what would they look like? If one of the children has children with a person from a family with a long history of straight noses, what types of noses would you expect the grandchildren to possess and in what proportions?

24 When plants that are pure-breeding for wrinkled seeds are crossed with plants that are pure-breeding for round seeds, the ratio of genotypes in the F_2 is 1:2:1 and the ratio of phenotypes is 3:1. Explain what causes the genotypic ratio to differ from the phenotypic ratio.

25 If a human male with blood group M has children with a female with blood group N, what blood groups would they possess? If one of the children has children with a person with blood group M, what blood groups could the grandchildren possess? Construct the crosses for each of these matings. List the genotypes and phenotypes that would be expected, and the probability of obtaining each genotype and phenotype.

26 A woman from a family with no history of haemophilia marries a man who is a haemophiliac. What is the probability that they will produce:

- **a** sons with normal blood clotting?
- **b** sons with haemophilia?
- **c** daughters who are carriers of haemophilia?
- **d** daughters who will be haemophiliacs?

27 Red–green colour blindness is a sex-linked characteristic. Under what circumstances would a couple produce daughters who all had normal vision and sons who were all colour blind? Describe the genotypes of both parents and all the children.

28 The first child born to a married couple with normal vision is a male with red–green colour blindness. Calculate the probability that their second child will also be colour blind. Give a clear explanation for your answer. Remember that red–green colour blindness is *X*-linked.

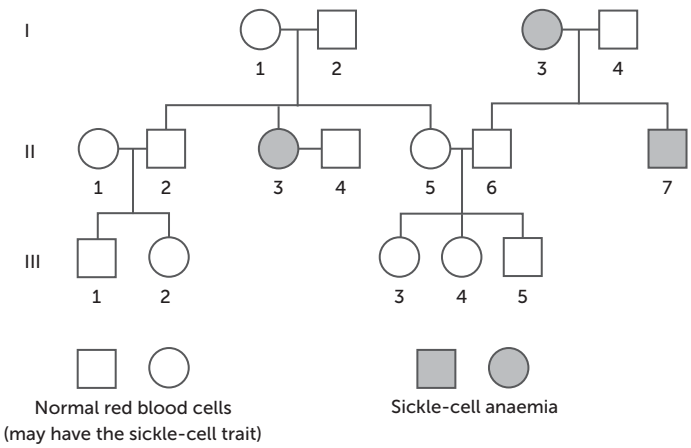
29 In the United States, about 6 in every 100 children whose parents are first cousins die before the age of 10 years. Where the parents are unrelated, the figure is about 2.5 in every 100. Can you suggest reasons for this big difference in mortality for the first 10 years of life?

30 **a** Chloe is Rh+ but her brother Jason is Rh–. Both Chloe's parents are Rh+. What is the probability that Chloe is a carrier for the recessive Rh allele?

- **b** Chloe married Mitchell and they had a daughter, Zara. What is the probability that Zara is Rh– if:
  - **i** Chloe is a carrier for the recessive allele?
  - **ii** Chloe is not a carrier for the recessive allele?

31 Examine the pedigree on the next page of families where some individuals have the sickle-cell allele.

9780170449090



a Which individuals have sickle-cell anaemia?

b Which individuals must have sickle-cell trait?

c Couple II 3 and II 4 are thinking of having a child. If you were a genetic counsellor, what advice would you give them regarding the possibility of having a child with sickle-cell anaemia or sickle-cell trait?

d None of the children of II 5 and II 6 have sickle-cell anaemia. Does this mean that the allele for sickle-cell anaemia no longer occurs in that branch of the family?

32 In October 2013, Irish police seized two children from Roma (Gypsy) families, claiming that they were too blonde to be the offspring of their dark-haired parents. DNA testing proved that the dark-haired couples were indeed their parents.

a How is it possible that two people with dark hair could have children with blonde hair?

b What is the probability that one of the couples could have another child with blonde hair?

Two blonde children returned to their Roma parents in Ireland as police are accused of racism

Extend

33 People with Huntington's disease often have children, even though their children will have at least a 50% chance of inheriting the disease. With such a high probability of passing the disease on, why do such people continue to have children?

34 With DNA profiling, genetically inherited diseases can be detected at an early age. Discuss the advantages of the early detection of a particular genetic disease.

35 Describe why it is impossible for parents who have the blood groups A and AB to produce children with blood group O.

36 Charlie Chaplin, a famous comedian of the silent screen, was taken to court in 1944 by a young starlet, Joan Barry. She claimed that Chaplin was the father of her child, and the court ruled in her favour. Blood group data was not admissible evidence at the time of that trial. However, if you were the judge, how would you have decided? The baby was blood group B, the mother A and Chaplin O. Give genetic reasons for your decision.

37 A woman has a brother with Duchenne muscular dystrophy. What information could be given to the woman about the

9780170449090

risk of her having a child with Duchenne muscular dystrophy?

38 In what situations would you be able to deduce a person's genotype by determining their phenotype? Give examples using ABO blood groups.

39 DNA profiling raises ethical questions. Discuss the ethical issues of the following situations.

a A wealthy grandparent suspects that a grandchild is not genetically related to her and plans to disinherit him if that is the case.

b A devoted fan is willing to pay a high price to purchase the genetic information of their favourite celebrity.

c A political party is interested in discovering and publicising any predispositions to disease that might render a candidate of the opposing party unsuitable for election.

9780170449090

INDEX

C

D

E

F

G

H

I

J

K

9780170449090

N

O

P

Q

R

S

T

U

V

W

X

Y

Z

9780170449090

nelson net.

ACE THIS SUBJECT

Want to go further with your learning?
Unlock your **NelsonNet** resources now.

TURN THE PAGE FOR YOUR ACCESS CODE!

Study **anywhere, anytime** with your downloadable **NelsonNetBook**

Go further with links to real-world information, animations and video tutorials

Worksheets help you **revise** important concepts

PLUS! GRAB YOUR FREE TIMETABLE

Scan this QR code to download and use it to help you plan your revision

www.nelsonnet.com.au

ISBN 978-0170449090